Massachusetts Birth Defects 1999

Bureau of Family and Community Health
Bureau of Health Statistics Research and Evaluation

Massachusetts Department of Public Health
November 2001

Massachusetts Birth Defects 1999

Jane Swift, Governor
Robert P. Gittens, Secretary, Executive Office of Health and Human Services
Howard K. Koh, MD, MPH, Commissioner, Massachusetts Department of Public Health
Deborah Klein Walker, Associate Commissioner for Programs and Prevention
Daniel J. Friedman, Assistant Commissioner, Bureau of Health Statistics, Research and Evaluation
Sally Fogerty, Assistant Commissioner, Bureau of Family and Community Health
Marlene Anderka, Director, Office of Statistics and Evaluation
Lisa D. Miller, Director, Massachusetts Center for Birth Defects Research and Prevention

Massachusetts Department of Public Health
617-624-5507

November, 2001

Acknowledgements

This report was prepared by the staff of the Massachusetts Center for Birth Defects Research and Prevention including: Marlene Anderka, Linda Baptiste, Elizabeth Bingay, Veronica Dornan, Neenah Estrella-Luna, Cathy Higgins, Annette Luc, Lisa Miller, Jean Murphy, Shannon Preston, and Theodora Wohler.

Data in this report have been collected through the efforts of the field staff of the Center for Birth Defects Research and Prevention including: Roberta Aucoin, Dorothy Cichonski, Daniel Sexton, Marie-Noel Westgate and Susan Winship.

We would like to acknowledge the following individuals for their time and commitment to supporting our efforts in improving the birth defects surveillance program.

Sonia Hernandez-Diaz, MD, PhD, Slone Epidemiology Unit, Boston University
Lewis Holmes, MD, Massachusetts General Hospital
Angela Lin, MD, Massachusetts General Hospital
Susan Littlefield, RN, Slone Epidemiology Unit, Boston University
Carol Louik, ScD, Slone Epidemiology Unit, Boston University
Allen Mitchell, MD, Slone Epidemiology Unit, Boston University
Martha Werler, ScD, Slone Epidemiology Unit, Boston University
Diego F. Wyszynski, MD, PhD, School of Medicine, Boston University

Antonia Blinn and staff
March of Dimes
Massachusetts Chapter

Elaine Trudeau and staff
Registry of Vital Records
Bureau of Health Statistics, Research and Evaluation
Massachusetts Department of Public Health

We would like to thank the Texas Birth Defects Monitoring Division, Texas Department of Health for their assistance. The layout of this report was modified from the Texas Birth Defects Registry Report of Birth Defects among 1996 and 1997 Deliveries.

We would like to acknowledge the following individuals for their support of the Massachusetts Center for Birth Defects Research and Prevention.

Larry Edmonds and staff
Birth Defects and Pediatric Genetics Branch
Division of Birth Defects, Child Development, Disability and Health
National Center for Environmental Health
Centers for Disease Control and Prevention

Funding for surveillance of 1999 birth defects in Massachusetts was provided by the Centers for Disease Control and Prevention via the Centers for Birth Defects Research and Prevention grant.

To obtain additional copies of this report, contact:
Massachusetts Department of Public Health
Bureau of Health Statistics, Research and Evaluation
Massachusetts Center for Birth Defects Research and Prevention
250 Washington Street
Boston, MA 02108
(617) 624-5507
This and other Department of Public Health publications and materials can be downloaded from the Internet at:
http://www.state.ma.us/dph/pubstats.htm

TABLE OF CONTENTS

EXECUTIVE SUMMARY *1*

Chapter 1: **INTRODUCTION**

Public Health Importance of Birth Defects *7*
Historical and Program Description *8*

Chapter 2: **METHODS**

Case Definition *11*
Data Collection *11*
Confidentiality *11*
Data Analysis *11*
Data Limitations *12*

Chapter 3: **PREVALENCE OF BIRTH DEFECTS**

Overall Prevalence of Birth Defects *15*
Table 1. Prevalence of Birth Defects 18
Table 2. Comparison of Massachusetts 1999 Observed Counts and Expected Counts 21
Figure 1. Distribution of Birth Defects by Defect Categories 22
Table 3. Most Common Defects Among Live Births And Stillbirths 23
Table 4. Single versus Multiple Defects Among Live Births and Stillbirths 24
Figure 2. Distribution of Selected Birth Defects by Single versus Multiple Defect Categories 28
Table 5. Prevalence of Birth Defects by Plurality of Live Births and Stillbirths 29
Figure 3. Plurality Distribution of All Live Births and Births Defect Cases 33
Table 6. Most Common Defects by Sex of Live Births and Stillbirths 34
Table 7. Prevalence of Birth Defects by Sex of Infants among Live Births and Stillbirths 35
Figure 4. Selected Birth Defects by Sex among Live Births and Stillbirths 39

Chapter 4: **PREVALENCE OF BIRTH DEFECTS BY MATERNAL AGE AND RACE/ETHNICITY**

Mother's Age and Mother's Race *41*
Table 8. Prevalence of Birth Defects by Mother's Age for Live Births 43
Table 9. Most Common Defects by Mother's Age 50
Figure 5. Distribution of Gastroschisis Among Mother's Age Groups 51
Figure 6. Distribution of Down Syndrome Among Mothers' Age Groups 52
Table 10. Prevalence of Birth Defects by Mother's Race /Hispanic Ethnicity for Live Births 53
Table 11. Most Common Defects by Mother's Race/Hispanic Ethnicity for Live Births 59
Table 12. Overall Distribution of Infants with Birth Defects by Mother's Ethnicity 60

Chapter 5: **PREVALENCE OF BIRTH DEFECTS BY REGION**

Prevalence of Birth Defects by Region *62*
Figure 7. Map of EOHHS Regions in Massachusetts 63
Table 13. Overall Distribution of Infants with Birth Defects by Region 64
Table 14. Most Common Defects by Region Among Live Births 65
Table 15. Prevalence of Birth Defects by Region 67

Chapter 6: **PREVALENCE OF BIRTH DEFECTS BY SEVERITY**

Prevalence of Birth Defects by Severity *70*
Table 16. Distribution of Birth Defects by Severity 72
Figure 8. Distribution of Cases with Birth Defects by Severity Groups 73

APPENDICES

Technical Notes *75*
Definitions *77*
1999 Populations Used in Calculating Rates *78*
Birth Defects Codes and Exclusions, Massachusetts: 1999 *79*
All ICD9-CM/BPA Codes with Counts-Live Births and Stillbirths *84*
Glossary of Birth Defect Terms *93*
References *98*

Executive Summary

Birth defects, when compared to other adverse birth outcomes, such as low birth weight and prematurity, are rare. These conditions, however, can be life threatening, lead to lifelong disability, and require costly medical care and multiple hospitalizations. The impact on the family may be enormous. Attempts to calculate costs for birth defects can severely underestimate the economic, emotional, and social burdens these families endure (Harris 1997).

According to the March of Dimes (MOD), one out of every twenty-eight infants is diagnosed with a birth defect (about 150,000 babies per year). The MOD definition of a birth defect is "an abnormality of structure, function or body metabolism (inborn error of body chemistry) present at birth that results in physical or mental disability, or is fatal" (March of Dimes 1997). MOD estimates there are more than 4,000 known birth defects. They are the leading cause of death in the first year of life and are the fifth leading cause of years of potential life lost in children.

The causes of birth defects are poorly understood. While certain genetic and environmental factors have been implicated with selected defects, at least 50% of major birth defects have unknown causes. Etiologic studies of birth defects are providing some answers. For instance, studies have shown that folic acid (vitamin B_9) may help to prevent defects of the brain and spinal cord known as neural tube defects.

Birth defects surveillance is a critical component of public health strategies to reduce the occurrence and impact of birth defects. The Centers for Disease Control and Prevention (CDC) has estimated that 3% to 5% of births have major structural birth defects based on ascertainment by an active surveillance system. Through surveillance, the Massachusetts Department of Public Health is able to detect the prevalence of birth defects, to investigate potential etiologic agents, to plan appropriate interventions, and to ensure services and appropriate care for children with special health care needs.

The primary focus of the Massachusetts active surveillance system[1] is identifying major structural birth defects. Other conditions monitored include selected genetic and chromosomal abnormalities. When an infant or fetus has a structural birth defect, some part of the body (internal or external) is missing or malformed. Structural birth defects are the major malformations monitored by most state surveillance systems and are the primary focus for the CDC birth defects system in Atlanta, Georgia. Inborn errors of metabolism are monitored by the state newborn screening program.

[1] Please see page 11 for the definition of an active surveillance system.

This report presents the first year of statewide data on the prevalence of major structural birth defects among live births and stillbirths to Massachusetts residents during calendar year 1999. Unless otherwise specified, this report uses the term "births" to mean live births plus stillbirths. A stillbirth is defined as the delivery of a fetus that is not alive, and is greater than or equal to 20 weeks gestational age, or weighs at least 350 grams.

Since 1999 represents the first full year of statewide birth defects surveillance, and birth defects are rare occurrences, interpretations of these data must be made with caution until a multiyear estimate establishes a stable, baseline rate.

Prevalence

The overall prevalence of birth defects among births to Massachusetts residents in 1999 is 111.8 per 10,000 live births. Five out of the ten most common defects are cardiovascular defects including patent ductus arteriosus, septal (atrial and ventricular) defects, coarctation of aorta, endocardial cushion defects, and pulmonary valve atresia and stenosis. Common non-cardiovascular defects include cleft lip (with and without cleft palate), cleft palate (without cleft lip), Down Syndrome, and obstructive genitourinary defects.

Among the 80,866 live births to Massachusetts residents in 1999, 875 have one or more birth defects. In addition, 29 stillbirths are identified as having a birth defect. Overall, 1.1% of births in the state have one or more birth defects.

This percentage is lower than the 3% to 5% estimated by CDC, perhaps due to the fact that Massachusetts currently has limited reporting. Counts for some defects (including anencephaly, spina bifida, and Trisomy 13) are less than 50% of the expected counts based on 1998 rates from the Metropolitan Atlanta Congenital Defects Program which includes cases among live births, stillbirths and elective terminations. These conditions may not be reported when they are prenatally diagnosed and the pregnancy is electively terminated. Stillbirths reported by birthing hospitals may not indicate whether there was a birth defect. Pathology reports offer detailed birth defects information but only on those stillbirths where an autopsy was performed. Other contributors to lower counts may be differences in defect criteria between the two surveillance systems and the fact that this was a start-up year for Massachusetts.

Single vs. Multiple Defects

Of all 904 birth defect cases (infants and stillborns), 55% have a single defect and 45% have multiple defects. Overall, cases are 22% less likely to have multiple defects. Anencephaly, cleft lip, gastroschisis, Hirschsprung Disease, and hypospadias appear more often as a single defect rather than with other defects. The majority of cardiovascular defects, limb reductions, microcephaly, hydrocephalus, and obstructive genitourinary defects appear more often in conjunction with other defects.

Plurality

Examining the birth defect rate by plurality is important to monitor since the number of multiple births is increasing over time in Massachusetts. The birth defect prevalence rate is 107.8 for singletons and 203.4 for multiple births (more than one infant) per 10,000 live births. Birth defects that are at least 25% more common in multiple births than in singletons include tethered cord, anophthalmia, esophageal atresia/tracheoesophageal fistula, Hirschsprung disease, and lower limb reduction defects.

Sex

The birth defect prevalence rate is 99.4 for females and 123.3 for males per 10,000 live births. While the majority of birth defects do not substantially differ by sex of the infant/fetus, some conditions are associated with sex. Common defects seen in both sexes include septal defects, Down Syndrome, cleft palate, cleft lip and pulmonary valve atresia and stenosis. The most common defects seen in males are cleft lip, craniosynostosis, gastroschisis, spina bifida, anotia/microtia, transposition of great arteries, and coarctation of aorta. The most common defects seen in females are Holoprosencephaly, hydrocephalus, hypoplastic left heart syndrome, small intestine atresia, and Trisomy 18.

Mother's Age

Examining birth defects by maternal age is important to monitor since the number of births to older mothers is increasing over time in Massachusetts. The prevalence of birth defects varies by mother's age group. Rates per 10,000 live births are 139.6 for less than 20 years old, 168.5 for 20-24 years old, 129.0 for 25-29 years old, 115.8 for 30-34 years old, and 175.5 for 35 years and older. As expected, there is a strong association of Down Syndrome with advanced mother's age. Women 35 years and older had a live birth Down Syndrome rate of 24.5 per 10,000 births. This rate is five times that of any other maternal age group. While results for other defects differ by age group, the small numbers from one year of surveillance are not sufficient for interpretation.

Mother's Race / Hispanic Ethnicity

The prevalence of birth defects varies by mother's race and Hispanic ethnicity. The rate per 10,000 live births is 122.5 for Hispanics, 107.8 for Blacks, 105.3 for Whites and 91.8 for Asians. The most common defects in Hispanics include septal defects, microcephaly, obstructive genitourinary defects, transposition of great arteries, Down syndrome, and cleft lip. In Blacks, the most common defects include septal defects, coarctation of aorta, Down Syndrome, hypospadias, microcephaly, and obstructive genitourinary defects. The most common defects in Whites include septal heart defects, Down Syndrome, cleft lip, and cleft palate. In Asians, the most common defects include cleft lip, hypospadias, gastroschisis, and omphalocele. Overall, birth defects prevalence also varies by detailed ethnicity; but due to the small numbers by specific defect, we need more years of data to examine ethnicity differences.

Region

The distribution and rates of birth defects were examined by the six regions of the state based on the Massachusetts Executive Office of Health and Human Services (EOHHS) designated areas. There is variation in both the overall rate and the most common birth defects by region. The prevalence rates per 10,000 live births are 147.3 in the West, 113.9 in the Central region, 111.6 in the Northeast, 97.0 in Metro West, 115.3 in Greater Boston and, 86.1 in the Southeast. The lower rate in the Southeast may be due to cases delivered in Rhode Island hospitals, where Massachusetts surveillance was not conducted in 1999. Differences in rates may also be due to differences in the demographics of the birth populations and in the way birth defects are diagnosed across regions. The most common defects (excluding patent ductus arteriosus) in all the regions include septal defects, Down Syndrome, cleft lip and cleft palate.

Severity

A severity scale was developed by the Center in collaboration with our partners at Boston University and the Massachusetts General Hospital. This scale was based on the usual outcome for a specific birth defect including its typical compatibility with survival, the need for immediate treatment, the need for long-term care, and the amenability of the defect to correction. A severity score was assigned to each case based on the most severe defect for that infant/fetus. Specific severity category definitions used in this report are as follows:

SEVERITY CATEGORIES	PERCENTAGE OF BIRTH DEFECTS CASES
Always Severe, Usually Incompatible with Life	3%
Severe Need Immediate Treatment Probable Long Term Needs	23%
Moderately Severe Usually Correctable	64%
Mild Minimal or No Correction Needed	10%

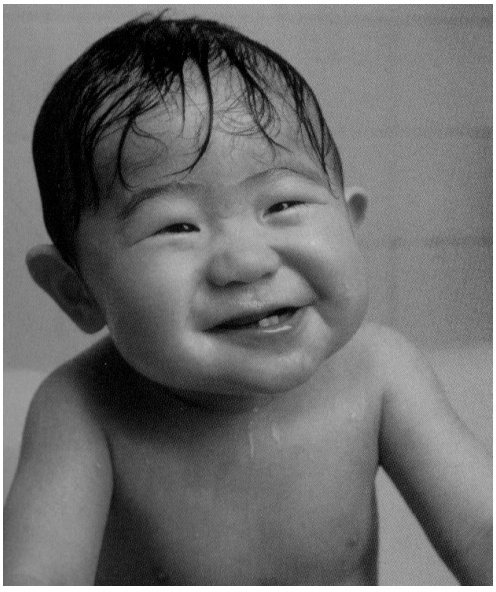
Three percent of cases with birth defects classified as “always severe” did not survive. This percentage is an underestimate of cases due to limited data. For example, Cragan at CDC has estimated that up to 80% of anencephaly cases and 50% of any neural tube defect may be electively terminated after prenatal diagnosis (Cragan 2000). Twenty-three percent of cases are affected with a

severe birth defect. These cases need intensive medical care and planning for continuing care and long-term disability. Moderately severe birth defects comprise 64% of the total cases, all of these will need medical follow up; many may require a number of surgeries and extensive treatment. Mild birth defects comprise 10% of the affected infants. These defects may or may not require corrective treatment.

Planning for children with special health care needs is essential to support affected infants and families. Coordination of birth defects data with maternal and child health programs helps to ensure services for identified children and to provide population-based information to inform program planning and prevention strategies.

Chapter 1

Introduction



Public Health Importance of Birth Defects

According to the March of Dimes (MOD), one out of every twenty-eight infants is diagnosed with a birth defect (about 150,000 babies per year nationwide). The MOD definition of a birth defect is, "an abnormality of structure, function or body metabolism (inborn error of body chemistry) present at birth that results in physical or mental disability, or is fatal" (MOD 1997). MOD estimates there are more than 4,000 known birth defects.

Birth defects are the fifth leading cause of years of potential life lost and a major cause of morbidity and mortality throughout childhood (Kochanek 1995, CDC 1989). Birth defects are also a leading cause of infant deaths representing 22% of overall infant mortality (less than one year of age) nationally. Among the 332 Massachusetts neonatal (less than 28 days of life) deaths occurring in 1999, 16.3% had one or more structural birth defects. More data are being collected to determine the contribution of birth defects to infant mortality in Massachusetts.

One Centers for Disease Control and Prevention (CDC) study showed that nearly 12% of pediatric hospitalizations in California and South Carolina combined were related to birth defects and genetic diseases. On average, these hospitalized children were about 3 years younger, stayed 3 days longer in a hospital, incurred 184% higher charges, and had a 4 1/2 times greater in-hospital mortality rate than children who were hospitalized for other reasons (Yoon 1997).

Estimates for lifetime costs of selected birth defects for several states (including Massachusetts) were generated from the average cost per child and the average prevalence rate of selected birth defects by the California Birth Defects Monitoring Program and the Metropolitan Atlanta Congenital Defects Program in 1992 (Harris 1997). Based on the 1992 estimates for Massachusetts adjusted for inflation, the combined lifetime costs of 12 major structural birth defects are an estimated $113 million dollars for 1999 (see Technical Notes).

A focus of Healthy People 2010 is to improve the health and well-being of women, infants, and children. The specific Healthy People 2010 objectives related to birth defects include: the reduction of fetal and infant death rates by 40%, the reduction of developmental disabilities rates by 50%, and the reduction of neural tube defects rates by 50%. Birth defects surveillance is a critical component of public health strategy to achieve these objectives.

Active surveillance and analysis of birth defects data enables the Department of Public Health to draw an accurate picture of the extent and occurrence of birth defects in the Commonwealth of Massachusetts. Data may also help to identify:

- Changes in birth defects rates over time that may indicate a change in environmental conditions and health of the population;
- Geographical areas with consistently high or unusual rates;
- Unusual clusters of birth defects;
- Families of affected children with birth defects who may benefit from services or who may be interested in participating in research studies; and

- Key data for preventive strategy planning by the Department of Public Health.

This report presents the first annual statewide data on the prevalence of birth defects among live births and stillbirths in Massachusetts during calendar year 1999. The primary focus of the surveillance system is the identification of structural birth defects. When an infant or stillborn has a structural birth defect, some part of the body (internal or external) is missing or malformed. The CDC has estimated that 3% to 5% of births have major structural birth defects based on ascertainment by an active surveillance system[2] (Edmonds 1997). Unless otherwise specified, this report uses the term "birth" to represent live births plus stillbirths.

Historical and Program Description

Widespread interest in birth defects in the U.S. was generated in the early 1960s, when an epidemic of limb reduction defects was associated with women's prenatal use of thalidomide (Edmonds 1997). Massachusetts passed legislation in 1963 that mandated birthing hospitals to report birth defects to the state Department of Public Health (MDPH) (M.G.L. Chapter 111, Section 67E).

During the 1970s, public health nurses maintained regular contact with birthing hospitals to promote reporting of birth defects to MDPH. During this time, however, the transport of acutely ill infants to non-birthing tertiary facilities (such as Children's Hospital) became more common and diagnoses frequently were not made until arrival at the non-birthing tertiary hospital. Such cases were increasingly not included in the birthing hospital report. This development led to under-reporting of cases in Massachusetts.

In 1984, a High-Risk Infant Identification System (HRIIS) was developed at MDPH. A form was filled out voluntarily by obstetric nurses in birth hospitals to capture defects associated with the risk of hearing loss. This system was cumbersome and found to understate the presence of birth defects, and was eventually phased out in the early nineties.

Administrative (passive) review of birth, death and fetal death certificates from the Registry of Vital Records and Statistics and the Uniform Hospital Discharge Data Set collected by the former Massachusetts Rate Setting Commission (now the Division of Health Care Finance and Policy) occurred during this time. Documentation of birth defects on birth certificates was inconsistent. As a result, the prevalence of these conditions was again underestimated.

In 1995, a pilot study matched 8,892 medical records from selected Massachusetts hospitals to birth certificates and records In the hospital discharge data. Of the 8,892 records, 365 (4%) had one major birth defect or at least 3 minor defects. Birth certificate data matched the medical record for 9% of the cases and hospital discharge data matched 55% of the cases (Bingay 1995). A three-year grant from

[2] Please see page 11 for an explanation of an active surveillance system.

CDC sparked further analysis of the administrative review data and revealed poor identification of cases in hospital discharge data and defect specific over-reporting and under-reporting of birth defects on the birth certificate.

In 1996, the CDC awarded five years of funding to MDPH to establish the Massachusetts Center for Birth Defects Research and Prevention (MCBDRP). The Center is a collaboration of MDPH with the Slone Epidemiology Unit at Boston University, and the Genetics and Teratology Department at Massachusetts General Hospital.

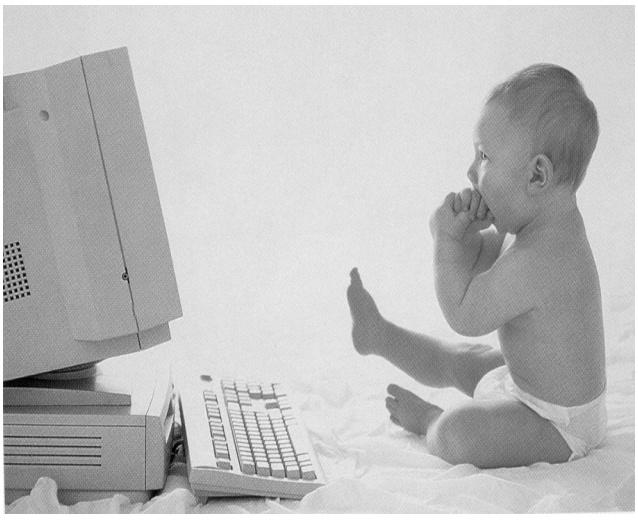
An active birth defects surveillance system involves trained personnel who validate passive reports of birth defects cases to the Department, and actively seek cases in hospitals and other health facilities. This approach provides more complete ascertainment of cases, more accurate data on cases, and more rapid reporting of cases. By working with hospital medical records departments, nurseries and neonatal intensive care units, the Center collected data on birth defect cases in the eastern part of the state from October 1997 to October 1998.

Since October 1998, MCBDRP surveillance program has been collecting cases statewide from 53 birth hospitals and Children's Hospital of Boston. This report of statewide 1999 data represents the first full year of active birth defects surveillance. Because this represents only one full year of surveillance data, and because birth defects are rare occurrences, interpretations of this data must be made with caution until a multiyear estimate establishes a stable, baseline rate.

The March of Dimes is sponsoring new legislation in Massachusetts to expand case ascertainment up to 3 years of age, and to expand reporting sources to include physicians, outpatient clinics, and genetic services. This update to the 1963 statute would make it possible for the Massachusetts surveillance program to obtain more accurate and complete birth defects data and to stabilize birth defects surveillance.

Chapter 2

Methods



Case Definition

This report presents data on selected birth defects occurring during the calendar year 1999 to Massachusetts residents. Cases met the following criteria:

- The infant was live born or, the fetus was stillborn with a gestational age greater than or equal to 20 weeks or with a weight of at least 350 grams.
- The infant or fetus had a structural birth defect that met diagnostic criteria (see Birth Defects Codes and Exclusions in Appendices).
- The diagnosis was made before the infant reached one year of age.

Data Collection

The Massachusetts Center for Birth Defects Research and Prevention uses active surveillance methods for population-based, statewide case ascertainment. Hospitals across the state submit monthly discharge lists with birth defect diagnoses to the Center. Nursery and neonatal intensive care liaisons phone in reports of birth defects. Abstractors review medical charts for each potential case. If the infant or fetus has a birth defect that meets the case definition criteria, detailed demographic and diagnostic information is recorded on a hospital reporting form. This information is entered into a confidential surveillance database for analysis.

Confidentiality

Great care is taken to protect the confidentiality of data. The Center has developed extensive procedures to guarantee the confidentiality of personal medical information and protect the privacy of families. These procedures uphold our ethical and legal obligations to safeguard confidentiality and fully comply with the strict requirements of state and federal laws.

Data Analysis

Counts for this report represent birth defects monitored in 1999. A defect may have occurred as a single event or with other defects. If the case had more than one defect within the same defect category, only one of these defects was counted in the category total. If the case had more than one defect in different defect categories, the case was represented in the total for each of these defect categories. Thus the counts in the defect categories presented in the prevalence tables cannot be added to obtain the total number of cases with birth defects.

The occurrence of birth defects is commonly reported as a prevalence. Prevalence is calculated as the number of birth defect cases born at a point in time per 10,000 live births. Prevalence tables include the number of cases found, the estimated prevalence rate per 10,000 live births, and the 95% confidence interval for that rate. The incidence (new cases) of birth defects (based upon the number of embryos conceived within a year) is not easily measured because both the total

number of conceptions that occur and the number of these conceptions resulting in a defect are not known (Sever 1996).

The confidence interval (CI) can be used to assess the magnitude and stability of a rate or ratio. The confidence interval (CI) for the rates in the tables is a range of values that has a 95% chance of including the underlying risk of an infant being born with a birth defect. Wide confidence intervals reflect the large variation due to small numbers (see Technical Notes).

Data Limitations

1. **Birth defect counts for this report are only for calendar year 1999. Due to the small numbers of birth defects, conclusions from these results are not valid until a multiyear estimate establishes a stable, baseline rate.**

2. The Massachusetts Center receives reports only from birthing hospitals and one non-birthing tertiary care center. Thus, defects that are not diagnosed at birth and that do not need hospitalization may be underreported (e.g., cardiac defects that are detected in an outpatient setting after the immediate newborn period).

3. Misclassification of birth defects may occur through coding errors or vague diagnoses. Quality control measures such as careful abstraction of the medical record minimize this error.

4. As medical diagnostic technology has improved, many prenatal and postnatal tests are now performed outside the traditional hospital setting. Prenatal diagnosis enables physicians to identify some birth defects well before the expected date of delivery, and offers women alternatives in the management of their affected pregnancies. These decisions have significant implications for monitoring birth defects. For example, it is estimated that up to 50% of all pregnancies affected with a neural tube defect may be discontinued and would thus not be included in hospital records (Cragan 2000). In addition, postnatal tests such as echocardiograms and ultrasounds may identify internal organ defects not diagnosed in the birthing hospital. Reporting of such postnatal results is not required by current law.

5. Stillbirths that are delivered prior to 20 weeks of gestation are not included in the case definition. It has been estimated that about 29% of birth defects cases are missed by not monitoring fetal demise prior to 20 weeks gestation (Forrester 1998, TBDR 2000).

6. Only diagnoses confirmed before one year of age are included. The frequency of diagnosed malformations can be higher among older children due to ‘hidden’ abnormalities such as kidney malformations or certain heart defects which are detected by accident or when a child is symptomatic (Holmes 1994). Another example, Fetal Alcohol Syndrome, may not be detected until developmental delays become evident when a child is much older.

7. Deliveries and diagnoses that occurred out of state are not included at this time. A review of 1998 Vital Statistics data indicates that approximately 1.3% of birth defects may be accounted among deliveries that occur across state borders. This will affect statistics for some regions of the state more than others. We are currently working with Rhode Island hospitals to capture affected deliveries for residents of Southeast Massachusetts who delivered in Rhode Island.

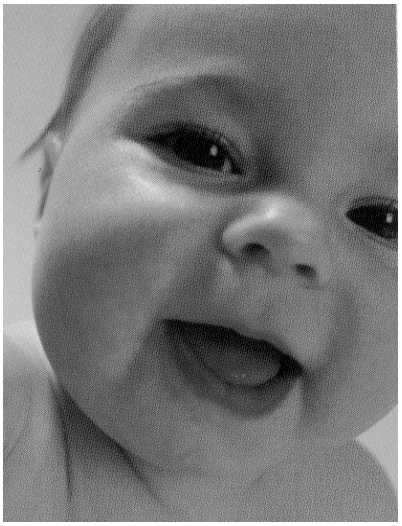
8. There are limitations in comparing data from the Metropolitan Atlanta Congenital Defects Program and the Massachusetts Center Birth Defects Monitoring Program. Factors such as differences in the demographics of the two populations, the environments in which they live, and the methods of surveillance conducted by the two programs may contribute to differences in the prevalence of birth defects.

Glossary

A glossary of birth defect terms is included in the appendices of this report.

Chapter 3

Prevalence of Birth Defects



Overall Prevalence of Birth Defects

Table 1 shows the prevalence of defects for all births and for live births only. Among the 80,866 live births to Massachusetts residents in 1999, 875 have one or more structural birth defects. In addition, 29 stillbirths are identified with a birth defect. A stillbirth is defined as the delivery of a fetus that is not alive, and is born with a gestational age greater than or equal to 20 weeks, or with a weight of at least 350 grams. Overall, 1.1% of births in the state have one or more birth defects. This is lower than the 3% to 5% estimated by CDC. This difference may be due to differences in criteria between the CDC and Massachusetts surveillance systems or in the reporting systems that the two surveillance systems have access to.

The Metropolitan Atlanta Congenital Defects Program (MACDP) collects defects on live births, stillbirths and elective terminations, using active surveillance methods in the Metropolitan Atlanta area. It is considered the "gold standard" of birth defects surveillance systems. Comparing expected birth defect counts from MACDP to the observed counts in Massachusetts helps to evaluate how our surveillance is doing in capturing cases.

Table 2 shows the comparison of 1999 observed counts for Massachusetts to expected counts generated by 1998 rates from MACDP. Expected counts are calculated from Atlanta rates which include live births, stillbirths, and elective terminations. Each birth defect specific rate from Atlanta was multiplied by the number of total 1999 Massachusetts births (80,866) to generate the expected numbers for that birth defect. A ratio of observed counts over expected counts (O/E) less than 1.0 indicates observed counts are less than expected counts. A ratio more than 1.0 indicates observed counts are greater than expected counts. Overall, the O/E ratio ranges between 0.8 to 1.2. If the confidence interval of the O/E ratio does not include 1.00, the observed counts from Massachusetts show a significant elevation or deficit compared to the Atlanta expected counts.

Massachusetts counts fell below 50% of the expected counts for anencephaly, spina bifida, and Trisomy 13. In general, these cases are prenatally diagnosed and, therefore may not be ascertained at a birthing hospital. CDC estimates that up to 50% of all neural tube defect cases may not be reported due to prenatal diagnosis and subsequent elective terminations (Cragan 2000). Spontaneous deliveries of stillbirths are reported by birthing hospitals but with limited information about the stillbirth on the mother's record. Pathology reports offer detailed birth defects information but typically only on those stillbirths where an autopsy was performed. Lower counts than expected may be due to differences in defect criteria between the two surveillance systems, and may also reflect the fact that this was a start-up year for Massachusetts.

The overall prevalence of reported birth defects in Massachusetts in 1999 was 111.8 per 10,000 live births. The majority of defects fell into cardiovascular (34%)

and musculoskeletal (26%) categories. Figure 1 shows the distribution of reported birth defects by defect categories.

The most common birth defect reported in Massachusetts is patent ductus arteriosus (PDA) at a rate of 12.7 per 10,000 live births. PDA is a cardiac blood vessel that remains open after birth (normally it spontaneously closes after birth). PDA varies in severity from mild to severe. Because the majority of these defects close and resolve spontaneously, we have excluded this defect in many of our analyses.

Table 3 shows the most common birth defects in the state. Five out of the ten most common defects are cardiovascular defects including patent ductus arteriosus, septal (atrial and ventricular) defects, coarctation of aorta, endocardial cushion defects, and pulmonary valve atresia and stenosis. Common non-cardiovascular defects include cleft lip, cleft palate, Down Syndrome, and obstructive genitourinary defects. Cardiovascular defects are the most commonly occurring birth defects in Massachusetts and nationally. They are also the largest defect contributing to infant deaths caused by birth defects (Petrini 1998).

Single vs. Multiple Defects

Table 4 shows the distribution of birth defects by whether they appear as a single diagnosis or in combination (multiple) with other defects.

Among birth defect cases, 55% have single defects and 45% have multiple defects. Figure 2 shows counts for selected birth defects by single and multiple defect categories.

Anencephaly, cleft lip, gastroschisis, Hirschsprung Disease, and hypospadias appear more often as a single defect rather than in combination with other defects. Limb reductions, microcephaly, hydrocephalus, and obstructive genitourinary defects appear more often with other defects. Overall, cardiovascular defects are eight times more likely to occur as one of multiple defects than as a single defect

Differences occur within defect categories (see Figure 2). Hypospadias occurs more frequently as a single diagnosis while obstructive genitourinary defects are more likely to occur in combination with other defects. A similar pattern appears with cleft lip and cleft palate.

Plurality

Table 5 depicts the distribution of birth defects by plurality. The overall prevalence is 107.8 for singletons and 203.4 for multiple births (more than one infant) per 10,000 live birth. Birth defects that are at least 25% more common in multiple births than in singletons include tethered cord, anophthalmia, esophageal atresia/tracheoesophageal fistula, Hirschsprung disease, and lower limb reduction defects. While multiple births comprise 4% of all births, they comprise 7% of all birth defects cases (see Figure 3). Examining birth defects by plurality is important

since the number of multiple births has been increasing over time in Massachusetts.

Sex

Table 6 shows the most common birth defects for females and males. The overall prevalence is 99.4 for females and 123.3 for males per 10,000 live births. While the majority of birth defects do not substantially differ by sex of the infant/fetus, some conditions are associated with sex. Common defects seen in both sexes include septal defects, Down Syndrome, cleft palate, cleft lip, and pulmonary valve atresia and stenosis. Figure 4 compares the prevalence of selected birth defects among males and females. The most common defects seen in males are cleft lip, craniosynostosis, gastroschisis, spina bifida, anotia/microtia, transposition of great arteries, and coarctation of aorta. The most common defects seen in females are holoprosencephaly, hydrocephalus, hypoplastic left heart syndrome, small intestine atresia, and Trisomy 18 (see Table 7).

Table 1 Prevalence of Birth Defects, Massachusetts: 1999

Defect[1]	Overall Count[2]	Rate[3]	95% Confidence Interval	Livebirth Count	Rate[3]	95% Confidence Interval
Central Nervous System						
Anencephaly	8	1.0	0.4 - 1.8	7	0.9	0.3 - 1.6
Spina Bifida	11	1.4	0.7 - 2.3	11	1.4	0.7 - 2.3
Hydrocephaly[4]	20	2.5	1.5 - 3.7	19	2.3	1.4 - 3.5
Encephalocele	4	0.5	0.1 - 1.1	4	0.5	0.1 - 1.1
Microcephaly	19	2.3	1.4 - 3.5	19	2.3	1.4 - 3.5
Holoprosencephaly	6	0.7	0.3 - 1.5	6	0.7	0.3 - 1.5
Tethered Cord	5	0.6	0.2 - 1.3	5	0.6	0.2 - 1.3
Total Central Nervous System Cases[5]	91			89		
Eye						
Anophthalmia/Microphthalmia	4	0.5	0.1 - 1.1	4	0.5	0.1 - 1.1
Congenital Cataract	11	1.4	0.7 - 2.3	11	1.4	0.7 - 2.3
Aniridia	1	0.1	0.0 - 0.5	1	0.1	0.0 - 0.5
Coloboma	6	0.7	0.3 - 1.5	6	0.7	0.3 - 1.5
Total Eye Cases[5]	24			24		
Ear						
Anotia/Microtia	12	1.5	0.8 - 2.4	12	1.5	0.8 - 2.4
Total Ear Cases[5]	13			13		
Cardiovascular/Respiratory						
Common Truncus/Aortic Septal Defects	1	0.1	0.0 - 0.5	1	0.1	0.0 - 0.5
Transposition of Great Arteries	21	2.6	1.6 - 3.8	21	2.6	1.6 - 3.8
Tetralogy of Fallot	24	3.0	1.9 - 4.3	24	3.0	1.9 - 4.3
Ventricular Septal Defect	71	8.8	6.9 - 10.9	68	8.4	6.5 - 10.5
Atrial Septal Defect	95	11.7	9.5 - 14.2	91	11.3	9.1 - 13.7
Endocardial Cushion Defect	25	3.1	2.0 - 4.4	24	3.0	1.9 - 4.3
Pulmonary Valve Atresia and Stenosis	34	4.2	2.9 - 5.7	33	4.1	2.8 - 5.6
Tricuspid Valve Atresia and Stenosis	7	0.9	0.3 - 1.6	7	0.9	0.3 - 1.6
Ebstein Anomaly	7	0.9	0.3 - 1.6	4	0.5	0.1 - 1.1
Aortic Valve Stenosis	12	1.5	0.8 - 2.4	12	1.5	0.8 - 2.4
Hypoplastic Left Heart Syndrome	14	1.7	0.9 - 2.8	12	1.5	0.8 - 2.4
Patent Ductus Arteriosus	103	12.7	10.4 - 15.3	103	12.7	10.4 - 15.3
Coarctation of Aorta	25	3.1	2.0 - 4.4	25	3.1	2.0 - 4.4

Table 1 Prevalence of Birth Defects, Massachusetts: 1999

Defect[1]	Overall Count[2]	Rate[3]	95% Confidence Interval	Livebirth Count	Rate[3]	95% Confidence Interval
Total Anomalous Pulmonary Venous Return	5	0.6	0.2 - 1.3	5	0.6	0.2 - 1.3
Partial Anomalous Pulmonary Venous Return	4	0.5	0.1 - 1.1	4	0.5	0.1 - 1.1
Single Ventricle	3	0.4	0.1 - 0.9	3	0.4	0.1 - 0.9
Situs Inversus/Heterotaxia	12	1.5	0.8 - 2.4	11	1.4	0.7 - 2.3
Total Cardiovascular/Respiratory Cases[5]	311			300		
Orofacial						
Cleft Palate without Cleft Lip	44	5.4	4.0 - 7.2	44	5.4	4.0 - 7.2
Cleft Lip with and without Cleft Palate	69	8.5	6.6 - 10.7	67	8.3	6.4 - 10.4
Choanal Atresia	2	0.2	0.0 - 0.7	2	0.2	0.0 - 0.7
Pierre Robin Sequence	15	1.9	1.0 - 2.9	15	1.9	1.0 - 2.9
Branchial Cleft/Sinus	8	1.0	0.4 - 1.8	8	1.0	0.4 - 1.8
Total Orofacial Cases[5]	126			124		
Gastrointestinal						
Esophageal Atresia/Tracheosophageal Fistula	23	2.8	1.8 - 4.1	20	2.5	1.5 - 3.7
Rectal and Large Intestinal Atresia/Stenosis	19	2.3	1.4 - 3.5	17	2.1	1.2 - 3.2
Hirschsprung Disease	10	1.2	0.6 - 2.1	10	1.2	0.6 - 2.1
Biliary Atresia	3	0.4	0.1 - 0.9	3	0.4	0.1 - 0.9
Small Intestinal Atresia	22	2.7	1.7 - 4.0	21	2.6	1.6 - 3.8
Total Gastrointestinal Cases[5]	91			87		
Genitourinary						
Renal Agenesis/Hypoplasia	4	0.5	0.1 - 1.1	3	0.4	0.1 - 0.9
Bladder Exstrophy	1	0.1	0.0 - 0.5	1	0.1	0.0 - 0.5
Obstructive Genitourinary Defect	42	5.2	3.7 - 6.9	42	5.2	3.7 - 6.9
Hypospadias	49	6.1	4.5 - 7.9	48	5.9	4.4 - 7.7
Ambiguous Genitalia	7	0.9	0.3 - 1.6	5	0.6	0.2 - 1.3
Total Genitourinary Cases[5]	124			116		
Musculoskeletal						
Reduction Deformity, Upper Limbs	15	1.9	1.0 - 2.9	14	1.7	0.9 - 2.8
Reduction Deformity, Lower limbs	8	1.0	0.4 - 1.8	6	0.7	0.3 - 1.5
Gastroschisis	14	1.7	0.9 - 2.8	14	1.7	0.9 - 2.8
Omphalocele	13	1.6	0.9 - 2.6	11	1.4	0.7 - 2.3
Diaphragmatic Hernia	17	2.1	1.2 - 3.2	16	2.0	1.1 - 3.1
Achondroplasia	2	0.2	0.0 - 0.7	2	0.2	0.0 - 0.7

Table 1 Prevalence of Birth Defects, Massachusetts: 1999

Defect[1]	Overall Count[2]	Rate[3]	95% Confidence Interval	Livebirth Count	Rate[3]	95% Confidence Interval
Thanatophoric Dysplasia	1	0.1	0.0 - 0.5	1	0.1	0.0 - 0.5
Osteogenesis Imperfecta	2	0.2	0.0 - 0.7	2	0.2	0.0 - 0.7
Craniosynostosis	14	1.7	0.9 - 2.8	14	1.7	0.9 - 2.8
Total Musculoskeletal Cases[5]	232			227		
Chromosomal/Syndromes						
Trisomy 13	2	0.2	0.0 - 0.7	2	0.2	0.0 - 0.7
Trisomy 21 (Down Syndrome)	67	8.3	6.4 - 10.4	65	8.0	6.2 - 10.1
Trisomy 18	16	2.0	1.1 - 3.1	11	1.4	0.7 - 2.3
Turner Syndrome	9	1.1	0.5 - 2.0	7	0.9	0.3 - 1.6
Klinefelter Syndrome	9	1.1	0.5 - 2.0	9	1.1	0.5 - 2.0
DiGeorge Syndrome	6	0.7	0.3 - 1.5	6	0.7	0.3 - 1.5
Total Chromosomal/Syndromes Cases[5]	157			145		
Other						
Amniotic Bands	6	0.7	0.3 - 1.5	4	0.5	0.1 - 1.1
Endocrine Anomalies	4	0.5	0.1 - 1.1	3	0.4	0.1 - 0.9
Epidermolysis Bullosa	2	0.2	0.0 - 0.7	2	0.2	0.0 - 0.7
X-linked Ichthyosis	2	0.2	0.0 - 0.7	2	0.2	0.0 - 0.7
Total Other Cases[5]	14			11		
Total Cases	904	111.8	104.6 - 119.2	875	108.2	101.2 - 115.5

[1] Cases can be included in more than one defect category. Cases are counted once within a defect category, therefore system totals will not equal sum of defect categories

[2] Overall count includes live births and stillbirths minus elective terminations and one adopted infant

[3] Rate per 10,000 livebirths

[4] Spina bifida includes cases with and without associated hydrocephaly

[5] Total categories in this table include other defects within the system category which have not been selected for presentation. See Appendix for complete list of defects included in these totals

Table 2 Comparison of Massachusetts 1999 Observed Counts and Expected Counts Based on Atlanta 1998 Rates per 10,000 Live Births[1]

	Atlanta 1998 Rates	Expected Counts[2]	Observed Counts[3]	O/E Ratio	95% Confidence Intervals[4]
CNS					
Anencephaly	4.00	32	8	0.25	0.09 - 0.83
Spina Bifida	5.34	43	11	0.25	0.09 - 0.84
Hydrocephaly without spina bifida	8.68	70	20	0.28	0.08 - 0.87
Encephalocele	1.11	9	4	0.45	0.04 - 1.03
Microcephaly	9.57	77	19	0.25	0.09 - 0.83
EYE					
Anophthalmia / Microphthalmia	2.23	18	4	0.22	0.10 - 0.80
Congenital Cataract	1.34	11	11	1.02	0.00 - 1.50
Aniridia	0.00	0	1		
EAR					
Anotia/Microtia	1.34	11	12	1.11	0.00 - 1.57
CARDIOVASCULAR					
Common Truncus	0.90	7	1	0.14	0.14 - 0.69
Transposition of Great Arteries	4.45	36	21	0.58	0.02 - 1.16
Tetralogy of Fallot	4.23	34	24	0.70	0.01 - 1.25
Endocardial Cushion Defect	4.23	34	25	0.73	0.01 - 1.28
Pulmonary Valve Atresia and Stenosis	5.56	45	34	0.76	0.00 - 1.30
Ebstein Anomaly	0.45	4	7	1.92	0.06 - 2.13
Coarctation of Aorta	5.79	47	25	0.53	0.02 - 1.11
Patent Ductus Arteriosus	35.61	288	103	0.36	0.06 - 0.95
Hypoplastic Left Heart Syndrome	3.34	27	14	0.52	0.03 - 1.10
Aortic Valve Stenosis	1.78	14	12	0.83	0.00 - 1.36
OROFACIAL					
Cleft Palate without Lip	6.23	50	44	0.87	0.00 - 1.39
Cleft Lip with and without Palate	10.91	88	69	0.78	0.00 - 1.32
Choanal Atresia	1.34	11	2	0.18	0.12 - 0.75
GI/GU					
Esophageal Atresia /Tracheoesphageal Fistula	0.89	7	23	3.20	0.25 - 2.91
Intestinal and Rectal Atresia/Stenosis	4.67	38	19	0.50	0.03 - 1.08
Hirschsprung Disease	2.00	16	10	0.62	0.01 - 1.18
Biliary Atresia	0.45	4	3	0.82	0.00 - 1.35
Bladder Exstrophy	0.45	0	1		
Obstructive Genitourinary	24.93	202	42	0.21	0.10 - 0.78
MUSCULOSKELETAL					
Upper Limb Reduction	4.01	32	15	0.46	0.03 - 1.05
Lower Limb Reduction	1.56	13	8	0.63	0.01 - 1.20
Gastroschisis	2.00	16	14	0.87	0.00 - 1.39
Omphalocele	2.45	20	13	0.66	0.01 - 1.22
Diaphragmatic Hernia	1.78	14	17	1.18	0.00 - 1.62
Amniotic Bands	0.89	7	6	0.83	0.00 - 1.36
CHROMOSOMAL					
Trisomy 13	1.34	11	2	0.18	0.12 - 0.75
Trisomy 21 (Down Sydrome)	12.69	103	67	0.65	0.01 - 1.21
Trisomy 18	2.67	22	16	0.74	0.01 - 1.29

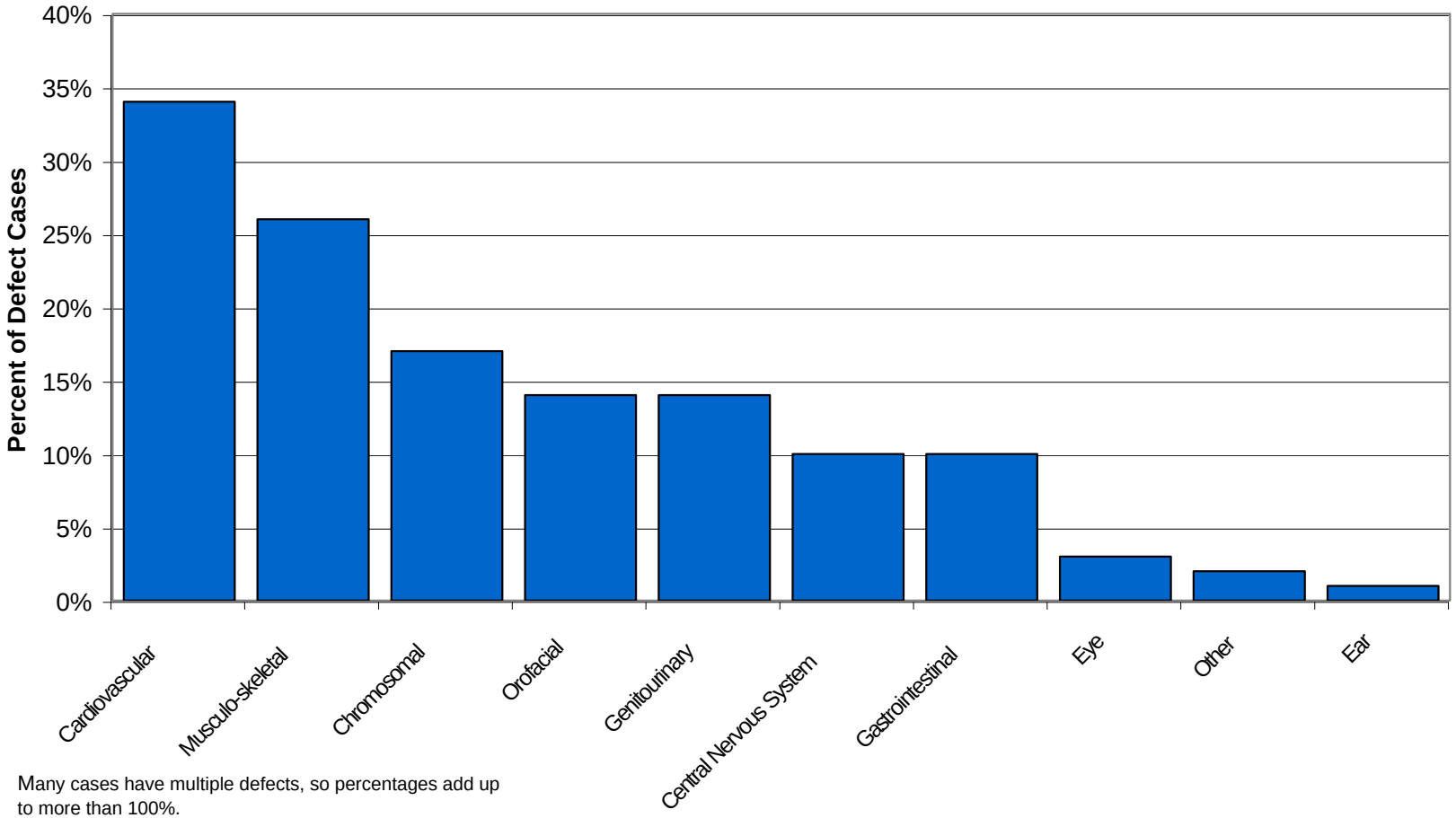
[1]Atlanta 1998 rates are provided by the Metropolitan Atlanta Congenital Defects Program

[2]Expected counts are calculated from Atlanta rates which include live births, stillbirths, and elective terminations. Each Atlanta rate was multiplied by the 1999 Massachusetts births total (80,866) to generate expected counts.

[3]Observed counts are the total live births and stillbirths counts for Massachusetts, 1999

[4] If the 95% CI range does not include 1.00, the observed counts are significantly elevated or deficient from the expected counts.

Figure 1 Distribution of Birth Defects by Defect Categories, Massachusetts: 1999



Many cases have multiple defects, so percentages add up to more than 100%.
Total Number of Defect Cases = 904

Table 3 Most Common Defects Among Live Births and Stillbirths, Massachusetts: 1999[1]

Defect	Count	Rate[2]	95% Confidence Intervals
Atrial Septal Defect	95	11.8	9.5 - 14.2
Ventricular Septal Defect	71	8.8	6.9 - 10.9
Cleft Lip with and without Cleft Palate	69	8.5	6.6 - 10.7
Trisomy 21 (Down Syndrome)	67	8.3	6.4 - 10.4
Hypospadias (2nd and 3rd Degree)	49	6.1	4.5 - 7.9
Cleft Palate without Cleft Lip	44	5.4	4.0 - 7.2
Obstructive Genitourinary Defect	42	5.2	3.7 - 6.9
Pulmonary Valve Atresia and Stenosis	34	4.2	2.9 - 5.7
Coarctation of the Aorta	25	3.1	2.0 - 4.4
Endocardial Cushion Defects	25	3.1	2.0 - 4.4

[1] Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

[2] Rate per 10,000 live births

Table 4 Single Versus Multiple Defects Among Live Births and Stillbirths, Massachusetts: 1999

Defect[1]	Cases With One Defect	Cases With Two or More Defects	Total Cases
Central Nervous System			
Anencephaly	7	1	8
Spina Bifida	7	4	11
Hydrocephaly[2]	4	16	20
Encephalocele	2	2	4
Microcephaly	5	14	19
Holoprosencephaly	0	6	6
Tethered Cord	2	3	5
Other Central Nervous System	10	26	36
Total Central Nervous System Cases[3]	37	72	109
Eye			
Anophthalmia/Microphthalmia	1	3	4
Congenital Cataract	5	6	11
Aniridia	1	0	1
Coloboma	2	4	6
Other Eye	1	3	4
Total Eye Cases[3]	10	16	26
Ear			0
Anotia/Microtia	4	8	12
Other Ear	1	4	5
Total Ear Cases[3]	5	12	17
Cardiovascular/Respiratory			
Common Truncus/Aortic Septal Defects	1	0	1
Transposition of Great Arteries	0	21	21
Tetralogy of Fallot	6	18	24
Ventricular Septal Defect	14	57	71
Atrial Septal Defect	20	75	95
Endocardial Cushion Defect	0	25	25
Pulmonary Valve Atresia and Stenosis	5	29	34
Tricuspid Valve Atresia and Stenosis	0	7	7
Ebstein Anomaly	1	6	7
Aortic Valve Stenosis	0	12	12
Hypoplastic Left Heart Syndrome	0	14	14

Table 4 Single Versus Multiple Defects Among Live Births and Stillbirths, Massachusetts: 1999

Defect[1]	Cases With One Defect	Cases With Two or More Defects	Total Cases
Patent Ductus Arteriosus	3	100	103
Coarctation of Aorta	3	22	25
Total Anomalous Pulmonary Venous Return	1	4	5
Partial Anomalous Pulmonary Venous Return	0	4	4
Single Ventricle	0	3	3
Situs Inversus/Heterotaxia	1	11	12
Other Cardiovascular/Respiratory	10	149	159
Total Cardiovascular/Respiratory Cases[3]	65	557	622
Orofacial			
Cleft Palate without Cleft Lip	18	26	44
Cleft Lip with and without Cleft Palate	58	11	69
Choanal Atresia	1	1	2
Pierre Robin Sequence	0	15	15
Branchial Cleft/Sinus	5	3	8
Other Orofacial	1	6	7
Total Orofacial Cases[3]	83	62	145
Gastrointestinal			
Esophageal Atresia/Tracheosophageal Fistula	11	12	23
Rectal and Large Intestinal Atresia/Stenosis	5	14	19
Hirschsprung Disease	8	2	10
Biliary Atresia	3	0	3
Small Intestinal Atresia	13	9	22
Other Gastrointestinal	6	16	22
Total Gastrointestinal Cases[3]	46	53	99
Genitourinary			
Renal Agenesis/Hypoplasia	1	3	4
Bladder Exstrophy	0	1	1
Obstructive Genitourinary Defect	7	35	42
Hypospadias	40	9	49
Ambiguous Genitalia	0	7	7
Other Genitourinary	12	28	40
Total Genitourinary Cases[3]	60	83	143

Table 4 Single Versus Multiple Defects Among Live Births and Stillbirths, Massachusetts: 1999

Defect[1]	Cases With One Defect	Cases With Two or More Defects	Total Cases
Musculoskeletal			
Reduction Deformity, Upper Limbs	3	12	15
Reduction Deformity, Lower limbs	2	6	8
Gastroschisis	11	3	14
Omphalocele	7	6	13
Diaphragmatic Hernia	10	7	17
Achondroplasia	2	0	2
Thanatophoric Dysplasia	0	1	1
Osteogenesis Imperfecta	1	1	2
Craniosynostosis	10	4	14
Other Musculoskeletal	78	88	166
Total Musculoskeletal Cases[3]	124	128	252
Chromosomal/Syndromes			
Trisomy 13	0	2	2
Trisomy 21 (Down Syndrome)	30	37	67
Trisomy 18	2	14	16
Turner Syndrome	8	1	9
Klinefelter Syndrome	7	2	9
DiGeorge Syndrome	0	6	6
Other Chromosomal/Syndromes	15	33	48
Total Chromosomal/Syndromes Cases[3]	62	95	157
Other			
Amniotic Bands	1	5	6
Endocrine Anomalies	0	4	4
Epidermolysis Bullosa	1	1	2
X-linked Ichthyosis	2	0	2
Total Other Cases[3]	4	10	14

Table 4 Single Versus Multiple Defects Among Live Births and Stillbirths, Massachusetts: 1999

Defect[1]	Cases With One Defect	Cases With Two or More Defects	Total Cases
Total Defects	497	1087	1584
Total Cases	497	407	904

[1] Cases can be in more than one defect category. For this table, infants are counted more than once within a category. System totals equal sum of defects.

[2] Spina bifida includes cases with and without associated hydrocephaly

[3] Total categories in this table include other defects within the system category. They are listed in "other" at the end of each system. See Appendix for complete list of defects included in these "other" categories.

Figure 2 Distribution of Selected Birth Defects by Single and Multiple Defect Categories, Massachusetts: 1999

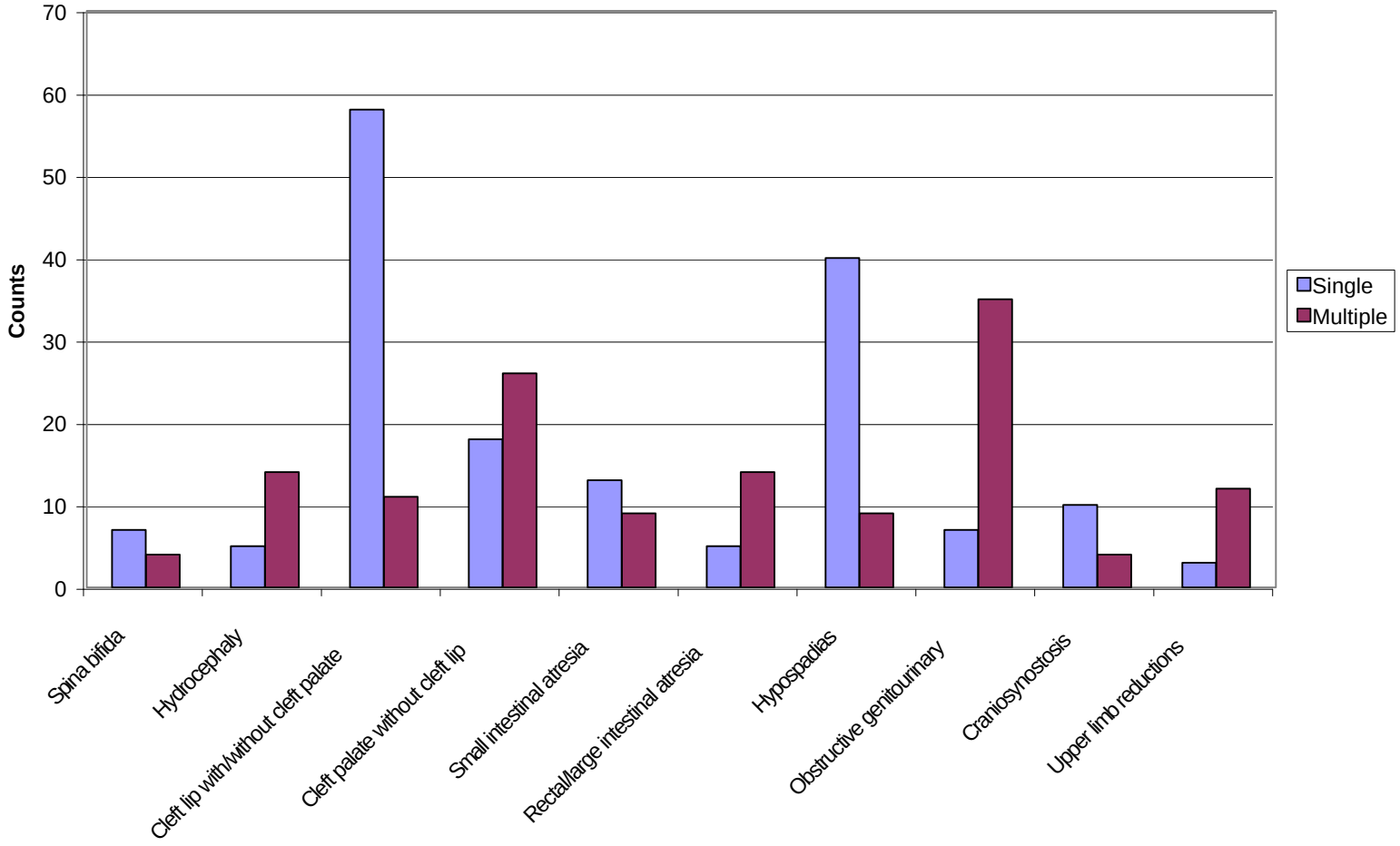


Table 5 (cont.) Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 1999

Defect[2]	Plurality	Cases	Rate[3]	95% Confidence Interval
CENTRAL NERVOUS SYSTEM				
Anencephaly	Singleton	6	0.8	0.3 - 1.5
	Multiple Birth	2	5.9	0.6 - 16.9
Spina Bifida	Singleton	10	1.3	0.6 - 2.2
	Multiple Birth	1	2.9	0.0 - 11.6
Hydrocephaly[4]	Singleton	17	2.2	1.3 - 3.4
	Multiple Birth	3	8.8	1.7 - 21.7
Encephalocele	Singleton	3	0.4	0.1 - 0.9
	Multiple Birth	1	2.9	0.0 - 11.6
Microcephaly	Singleton	18	2.3	1.4 - 3.5
	Multiple Birth	1	2.9	0.0 - 11.6
Holoprosencephaly	Singleton	6	0.8	0.3 - 1.5
	Multiple Birth	0	0.0	
Tethered Cord	Singleton	2	0.3	0.0 - 0.7
	Multiple Birth	3	8.8	1.7 - 21.7
Total Central Nervous System Cases[5]		91		
EYE				
Anophthalmia/Microphthalmia	Singleton	3	0.4	0.1 - 0.9
	Multiple Birth	1	2.9	0.0 - 11.6
Congenital Cataract	Singleton	11	1.4	0.7 - 2.4
	Multiple Birth	0	0.0	
Aniridia	Singleton	1	0.1	0.0 - 0.5
	Multiple Birth	0	0.0	
Coloboma	Singleton	5	0.6	0.2 - 1.3
	Multiple Birth	1	2.9	0.0 - 11.6
Total Eye Cases[5]		24		
EAR				
Anotia/Microtia	Singleton	11	1.4	0.7 - 2.4
	Multiple Birth	1	2.9	0.0 - 11.6
Total Ear Cases[5]		13		
CARDIOVASCULAR/RESPIRATORY				
Common Truncus/Aortic Septal Defects	Singleton	1	0.1	0.0 - 0.5
	Multiple Birth	0	0.0	
Transposition of Great Arteries	Singleton	20	2.6	1.6 - 3.8
	Multiple Birth	1	2.9	0.0 - 11.6
Tetralogy of Fallot	Singleton	23	3.0	1.9 - 4.3
	Multiple Birth	1	2.9	0.0 - 11.6
Ventricular Septal Defect	Singleton	66	8.5	6.6 - 10.7
	Multiple Birth	5	14.7	4.6 - 30.5
Atrial Septal Defect	Singleton	85	11.0	8.8 - 13.4
	Multiple Birth	10	29.5	14.0 - 50.6
Endocardial Cushion Defect	Singleton	23	3.0	1.9 - 4.3
	Multiple Birth	2	5.9	0.6 - 16.9
Pulmonary Valve Atresia and Stenosis	Singleton	32	4.1	2.8 - 5.7
	Multiple Birth	2	5.9	0.6 - 16.9

Table 5 (cont.) Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 1999

Defect[2]	Plurality	Cases	Rate[3]	95% Confidence Interval
Tricuspid Valve Atresia and Stenosis	Singleton	6	0.8	0.3 - 1.5
	Multiple Birth	1	2.9	0.0 - 11.6
Ebstein Anomaly	Singleton	6	0.8	0.3 - 1.5
	Multiple Birth	1	2.9	0.0 - 11.6
Aortic Valve Stenosis	Singleton	12	1.5	0.8 - 2.5
	Multiple Birth	0	0.0	
Hypoplastic Left Heart Syndrome	Singleton	14	1.8	1.0 - 2.9
	Multiple Birth	0	0.0	
Patent Ductus Arteriosus	Singleton	95	12.3	9.9 - 14.9
	Multiple Birth	8	23.6	10.1 - 42.7
Coarctation of Aorta	Singleton	21	2.7	1.7 - 4.0
	Multiple Birth	4	11.8	3.1 - 26.2
Total Anomalous Pulmonary Venous Return	Singleton	4	0.5	0.1 - 1.1
	Multiple Birth	1	2.9	0.0 - 11.6
Partial Anomalous Pulmonary Venous Return	Singleton	4	0.5	0.1 - 1.1
	Multiple Birth	0	0.0	
Single Ventricle	Singleton	3	0.4	0.1 - 0.9
	Multiple Birth	0	0.0	
Situs Inversus/Heterotaxia	Singleton	10	1.3	0.6 - 2.2
	Multiple Birth	2	5.9	0.6 - 16.9
Total Cardiovascular/Respiratory Cases[5]		311		
OROFACIAL				
Cleft Palate without Cleft Lip	Singleton	42	5.4	3.9 - 7.2
	Multiple Birth	2	5.9	0.6 - 16.9
Cleft Lip with and without Cleft Palate	Singleton	66	8.5	6.6 - 10.7
	Multiple Birth	3	8.8	1.7 - 21.7
Choanal Atresia	Singleton	2	0.3	0.0 - 0.7
	Multiple Birth	0	0.0	
Pierre Robin Sequence	Singleton	14	1.8	1.0 - 2.9
	Multiple Birth	1	2.9	0.0 - 11.6
Brachial Cleft/Sinus	Singleton	8	1.0	0.4 - 1.9
	Multiple Birth	0	0.0	
Total Orofacial Cases[5]		126		
GASTROINTESTINAL				
Esophageal Atresia/Tracheoesophageal Fistula	Singleton	17	2.2	1.3 - 3.4
	Multiple Birth	6	17.7	6.4 - 34.7
Rectal and Large Intestinal Atresia/Stenosis	Singleton	16	2.1	1.2 - 3.2
	Multiple Birth	3	8.8	1.7 - 21.7
Hirschsprung Disease	Singleton	8	1.0	0.4 - 1.9
	Multiple Birth	2	5.9	0.6 - 16.9
Biliary Atresia	Singleton	3	0.4	0.1 - 0.9
	Multiple Birth	0	0.0	
Small Intestinal Atresia	Singleton	20	2.6	1.6 - 3.8
	Multiple Birth	2	5.9	0.6 - 16.9
Total Gastrointestinal Cases[5]		91		

Table 5 (cont.) Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 1999

Defect[2]	Plurality	Cases	Rate[3]	95% Confidence Interval
GENITOURINARY				
Renal Agenesis/Hypoplasia	Singleton	4	0.5	0.1 - 1.1
	Multiple Birth	0	0.0	
Bladder Exstrophy	Singleton	1	0.1	0.0 - 0.5
	Multiple Birth	0	0.0	
Obstructive Genitourinary Defect	Singleton	39	5.0	3.6 - 6.7
	Multiple Birth	3	8.8	1.7 - 21.7
Hypospadias	Singleton	43	5.6	4.0 - 7.3
	Multiple Birth	6	17.7	6.4 - 34.7
Ambiguous Genitalia	Singleton	7	0.9	0.4 - 1.7
	Multiple Birth	0	0.0	
Total Genitourinary Cases[5]		124		
MUSCULOSKELETAL				
Reduction Deformity, Upper Limbs	Singleton	13	1.7	0.9 - 2.7
	Multiple Birth	2	5.9	0.6 - 16.9
Reduction Deformity, Lower Limbs	Singleton	6	0.8	0.3 - 1.5
	Multiple Birth	2	5.9	0.6 - 16.9
Gastroschisis	Singleton	14	1.8	1.0 - 2.9
	Multiple Birth	0	0.0	
Omphalocele	Singleton	12	1.5	0.8 - 2.5
	Multiple Birth	1	2.9	0.0 - 11.6
Diaphragmatic Hernia	Singleton	16	2.1	1.2 - 3.2
	Multiple Birth	1	2.9	0.0 - 11.6
Achondroplasia	Singleton	2	0.3	0.0 - 0.7
	Multiple Birth	0	0.0	
Thanatophoric Dysplasia	Singleton	1	0.1	0.0 - 0.5
	Multiple Birth	0	0.0	
Osteogenesis Imperfecta	Singleton	2	0.3	0.0 - 0.7
	Multiple Birth	0	0.0	
Craniosynostosis	Singleton	13	1.7	0.9 - 2.7
	Multiple Birth	1	2.9	0.0 - 11.6
Total Musculoskeletal Cases[5]		232		
CHROMOSOMAL/SYNDROMES				
Trisomy 13	Singleton	2	0.3	0.0 - 0.7
	Multiple Birth	0	0.0	
Trisomy 21 (Down Syndrome)	Singleton	65	8.4	6.5 - 10.6
	Multiple Birth	2	5.9	0.6 - 16.9
Trisomy 18	Singleton	16	2.1	1.2 - 3.2
	Multiple Birth	0	0.0	
Turner Syndrome	Singleton	9	1.2	0.5 - 2.0
	Multiple Birth	0	0.0	
Klinefelter Syndrome	Singleton	9	1.2	0.5 - 2.0
	Multiple Birth	0	0.0	
Total Chromosomal Cases[5]		109		

Table 5 (cont.) Prevalence of Birth Defects by Plurality[1] of Live Births and Stillbirths, Massachusetts: 1999

Defect[2]	Plurality	Cases	Rate[3]	95% Confidence Interval
OTHER				
Amniotic Bands	Singleton	6	0.8	0.3 - 1.5
	Multiple Birth	0	0.0	
Endocrine Anomalies	Singleton	4	0.5	0.1 - 1.1
	Multiple Birth	0	0.0	
Epidermolysis Bullosa	Singleton	2	0.3	0.0 - 0.7
	Multiple Birth	0	0.0	
X-linked Ichthyosis	Singleton	2	0.3	0.0 - 0.7
	Multiple Birth	0	0.0	
Total Other Cases[5]		14		
Total Cases		904		

[1] Plurality is the number of births to a woman from the same pregnancy. A singleton is the birth of one infant. Multiple birth represents more than one infant.

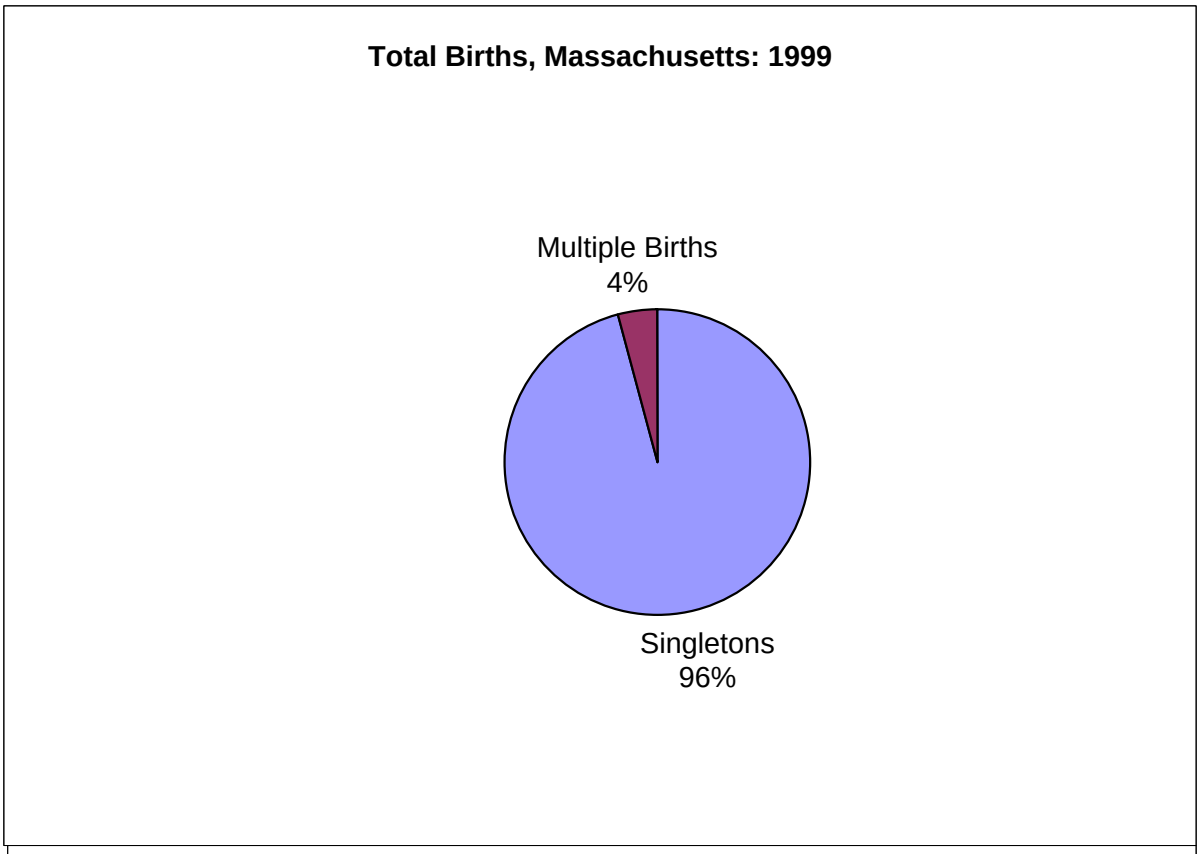
[2] Cases can be included in more than one defect category. Cases are counted once within a defect category, therefore system totals will not equal sum of defect categories

[3] Rate per 10,000 live births

[4] Spina bifida includes cases with and without associated hydrocephaly

[5] Total categories in this table include other defects within the system category which have not been selected for presentation. See Appendix for complete list of defects included in these totals.

Figure 3 Plurality Distribution of All Live Births and Birth Defect Cases



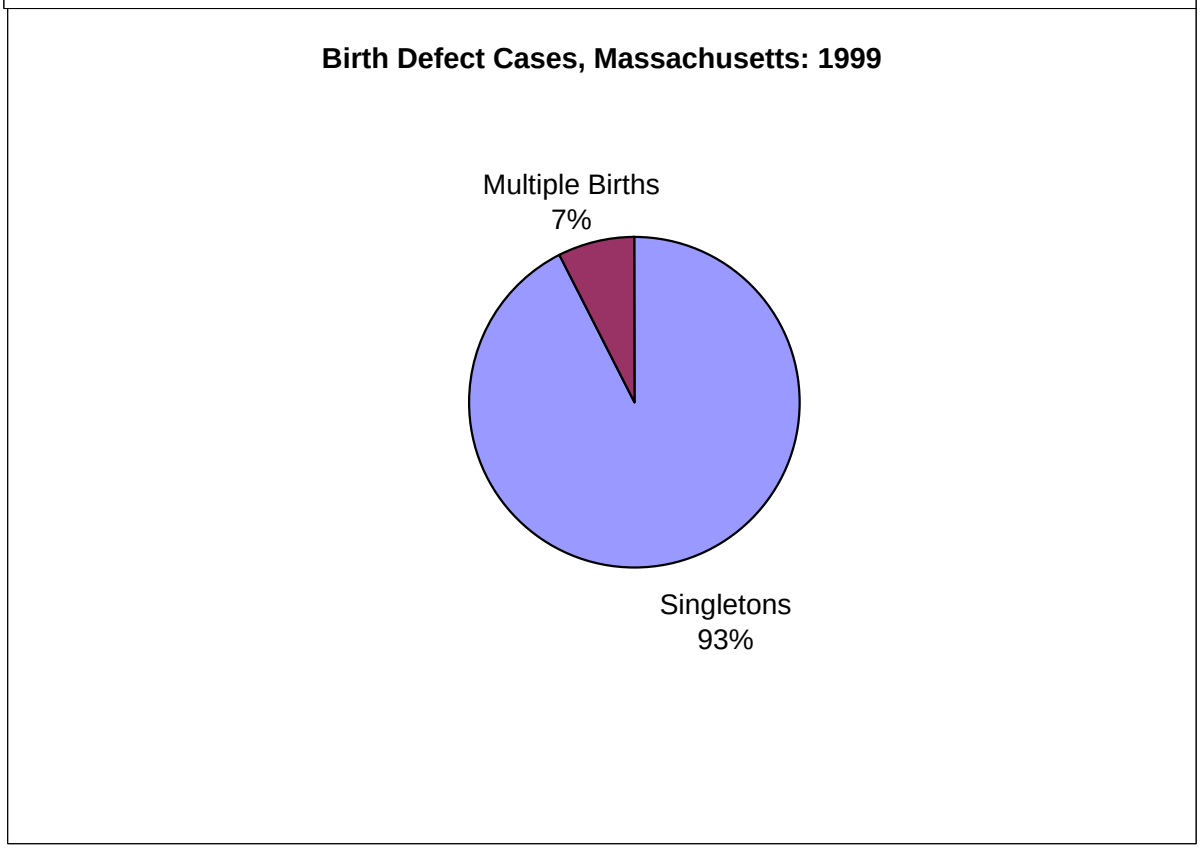


Table 6 Most Common Defects by Sex of Live Births and Stillbirths[1], Massachusetts: 1999

	Defect	Count	Rate[2]	95 % Confidence Intervals
Female				
	Atrial Septal Defect	50	12.7	9.4 - 16.4
	Trisomy 21 (Down Syndrome)	38	9.6	6.8 - 12.9
	Ventricular Septal Defect	37	9.4	6.6 - 12.7
	Cleft Lip with and without Cleft Palate	24	6.1	3.9 - 8.8
	Cleft Palate without Cleft Lip	22	5.6	3.5 - 8.2
	Pulmonary Valve Atresia and Stenosis	21	5.3	3.3 - 7.8
	Small Intestinal Atresia	18	4.6	2.7 - 6.9
	Endocardial Cushion Defect	15	3.8	2.1 - 6.0
	Hydrocephaly	13	3.3	1.7 - 5.3
	Tetralogy of Fallot	12	3.0	1.6 - 5.0
Male				
	Hypospadias (2nd and 3rd Degree)	49	11.8	8.7 - 15.4
	Atrial Septal Defect	45	10.9	7.9 - 14.3
	Cleft Lip with and without Cleft Palate	44	10.6	7.7 - 14.0
	Ventricular Septal Defect	34	8.2	5.7 - 11.2
	Obstructive Genitourinary Defect	33	8.0	5.5 - 10.9
	Trisomy 21 (Down Syndrome)	29	7.0	4.7 - 9.8
	Coarctation of Aorta	22	5.3	3.3 - 7.8
	Cleft Palate without Cleft Lip	22	5.3	3.3 - 7.8
	Transposition of Great Arteries	17	4.1	2.4 - 6.3
	Rectal and Large Intestinal Atresia/Stenosis	14	3.4	1.8 - 5.4

[1] Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

[2] Rate per 10,000 live births

Table 7 Prevalence of Birth Defects by Sex of Infant Among Live Births and Stillbirths, Massachusetts: 1999

Defect[1]	Sex[2]	Cases	Rate[3]	95% Confidence Interval
CENTRAL NERVOUS SYSTEM				
Anencephaly	Male	3	0.7	0.1 - 1.8
	Female	5	1.3	0.4 - 2.6
Spina Bifida	Male	9	2.2	1.0 - 3.8
	Female	2	0.5	0.0 - 1.5
Hydrocephaly[4]	Male	7	1.7	0.7 - 3.2
	Female	13	3.3	1.7 - 5.3
Encephalocele	Male	1	0.2	0.0 - 0.9
	Female	3	0.8	0.1 - 1.9
Microcephaly	Male	8	1.9	0.8 - 3.5
	Female	11	2.8	1.4 - 4.7
Holoprosencephaly	Male	2	0.5	0.0 - 1.4
	Female	4	1.0	0.3 - 2.3
Tethered Cord	Male	5	1.2	0.4 - 2.5
	Female	0	0.0	
Total Central Nervous System Cases[5]		91		
EYE				
Anophthalmia/Microphthalmia	Male	4	1.0	0.3 - 2.1
	Female	0	0.0	
Congenital Cataract	Male	5	1.2	0.4 - 2.5
	Female	6	1.5	0.5 - 3.0
Aniridia	Male	1	0.2	0.0 - 0.9
	Female	0	0.0	
Coloboma	Male	3	0.7	0.1 - 1.8
	Female	3	0.8	0.1 - 1.9
Total Eye Cases[5]		24		
EAR				
Anotia/Microtia	Male	9	2.2	1.0 - 3.8
	Female	3	0.8	0.1 - 1.9
Total Ear Cases[5]		13		
CARDIOVASCULAR/RESPIRATORY				
Common Truncus/Aortic Septal Defects	Male	1	0.2	0.0 - 0.9
	Female	0	0.0	
Transposition of Great Arteries	Male	17	4.1	2.4 - 6.3
	Female	4	1.0	0.3 - 2.3
Tetralogy of Fallot	Male	12	2.9	1.5 - 4.8
	Female	12	3.0	1.6 - 5.0
Ventricular Septal Defect	Male	34	8.2	5.7 - 11.2
	Female	37	9.4	6.6 - 12.7
Atrial Septal Defect	Male	45	10.9	7.9 - 14.3
	Female	50	12.7	9.4 - 16.4
Endocardial Cushion Defect	Male	10	2.4	1.1 - 4.1
	Female	15	3.8	2.1 - 6.0
Pulmonary Valve Atresia and Stenosis	Male	13	3.1	1.7 - 5.1
	Female	21	5.3	3.3 - 7.8

Table 7 Prevalence of Birth Defects by Sex of Infant Among Live Births and Stillbirths, Massachusetts: 1999

Defect[1]	Sex[2]	Cases	Rate[3]	95% Confidence Interval
Tricuspid Valve Atresia and Stenosis	Male	4	1.0	0.3 - 2.1
	Female	3	0.8	0.1 - 1.9
Ebstein Anomaly	Male	3	0.7	0.1 - 1.8
	Female	4	1.0	0.3 - 2.3
Aortic Valve Stenosis	Male	5	1.2	0.4 - 2.5
	Female	7	1.8	0.7 - 3.3
Hypoplastic Left Heart Syndrome	Male	5	1.2	0.4 - 2.5
	Female	9	2.3	1.0 - 4.0
Patent Ductus Arteriosus	Male	66	15.9	12.3 - 20.0
	Female	37	9.4	6.6 - 12.7
Coarctation of Aorta	Male	22	5.3	3.3 - 7.8
	Female	3	0.8	0.1 - 1.9
Total Anomalous Pulmonary Venous Return	Male	4	1.0	0.3 - 2.1
	Female	1	0.3	0.0 - 1.0
Partial Anomalous Pulmonary Venous Return	Male	1	0.2	0.0 - 0.9
	Female	3	0.8	0.1 - 1.9
Single Ventricle	Male	2	0.5	0.0 - 1.4
	Female	1	0.3	0.0 - 1.0
Situs Inversus/Heterotaxia	Male	4	1.0	0.3 - 2.1
	Female	8	2.0	0.9 - 3.7
Total Cardiovascular/Respiratory Cases[5]		311		
OROFACIAL				
Cleft Palate without Cleft Lip	Male	22	5.3	3.3 - 7.8
	Female	22	5.6	3.5 - 8.2
Cleft Lip with and without Cleft Palate	Male	44	10.6	7.7 - 14.0
	Female	24	6.1	3.9 - 8.8
Choanal Atresia	Male	2	0.5	0.0 - 1.4
	Female	0	0.0	
Pierre Robin Sequence	Male	6	1.4	0.5 - 2.8
	Female	9	2.3	1.0 - 4.0
Brachial Cleft/Sinus	Male	4	1.0	0.3 - 2.1
	Female	4	1.0	0.3 - 2.3
Total Orofacial Cases[5]		126		
GASTROINTESTINAL				
Esophageal Atresia/Tracheoesophageal Fistula	Male	11	2.7	1.3 - 4.5
	Female	11	2.8	1.4 - 4.7
Rectal and Large Intestinal Atresia/Stenosis	Male	14	3.4	1.8 - 5.4
	Female	5	1.3	0.4 - 2.6
Hirschsprung Disease	Male	5	1.2	0.4 - 2.5
	Female	5	1.3	0.4 - 2.6
Biliary Atresia	Male	0	0.0	
	Female	3	0.8	0.1 - 1.9
Small Intestinal Atresia	Male	4	1.0	0.3 - 2.1
	Female	18	4.6	2.7 - 6.9
Total Gastrointestinal Cases[5]		91		

Table 7 Prevalence of Birth Defects by Sex of Infant Among Live Births and Stillbirths, Massachusetts: 1999

Defect[1]	Sex[2]	Cases	Rate[3]	95% Confidence Interval
GENITOURINARY				
Renal Agenesis/Hypoplasia	Male	1	0.2	0.0 - 0.9
	Female	3	0.8	0.1 - 1.9
Bladder Exstrophy	Male	1	0.2	0.0 - 0.9
	Female	0	0.0	
Obstructive Genitourinary Defect	Male	33	8.0	5.5 - 10.9
	Female	9	2.3	1.0 - 4.0
Hypospadias	Male	49	11.8	8.7 - 15.4
	Female	0	0.0	
Ambiguous Genitalia	Male	2	0.5	0.0 - 1.4
	Female	4	1.0	0.3 - 2.3
Total Genitourinary Cases[5]		124		
MUSCULOSKELETAL				
Reduction Deformity, Upper Limbs	Male	9	2.2	1.0 - 3.8
	Female	6	1.5	0.5 - 3.0
Reduction Deformity, Lower Limbs	Male	3	0.7	0.1 - 1.8
	Female	5	1.3	0.4 - 2.6
Gastroschisis	Male	10	2.4	1.1 - 4.1
	Female	4	1.0	0.3 - 2.3
Omphalocele	Male	6	1.4	0.5 - 2.8
	Female	6	1.5	0.5 - 3.0
Diaphragmatic Hernia	Male	10	2.4	1.1 - 4.1
	Female	6	1.5	0.5 - 3.0
Achondroplasia	Male	2	0.5	0.0 - 1.4
	Female	0	0.0	
Thanatophoric Dysplasia	Male	1	0.2	0.0 - 0.9
	Female	0	0.0	
Osteogenesis Imperfecta	Male	0	0.0	
	Female	2	0.5	0.0 - 1.5
Craniosynostosis	Male	10	2.4	1.1 - 4.1
	Female	4	1.0	0.3 - 2.3
Total Musculoskeletal Cases[5]		232		
CHROMOSOMAL/SYNDROMES				
Trisomy 13	Male	1	0.2	0.0 - 0.9
	Female	1	0.3	0.0 - 1.0
Trisomy 21 (Down Syndrome)	Male	29	7.0	4.7 - 9.8
	Female	38	9.6	6.8 - 12.9
Trisomy 18	Male	4	1.0	0.3 - 2.1
	Female	11	2.8	1.4 - 4.7
Turner Syndrome	Male	0	0.0	
	Female	9	2.3	1.0 - 4.0
Klinefelter Syndrome	Male	9	2.2	1.0 - 3.8
	Female	0	0.0	
DiGeorge Syndrome	Male	4	1.0	0.3 - 2.1
	Female	2	0.5	0.0 - 1.5
Total Chromosomal/Syndromes Cases[5]		157		

Table 7 Prevalence of Birth Defects by Sex of Infant Among Live Births and Stillbirths, Massachusetts: 1999

Defect[1]	Sex[2]	Cases	Rate[3]	95% Confidence Interval
OTHER				
Amniotic Bands	Male	3	0.7	0.1 - 1.8
	Female	3	0.8	0.1 - 1.9
Endocrine Anomalies	Male	2	0.5	0.0 - 1.4
	Female	2	0.5	0.0 - 1.5
Epidermolysis Bullosa	Male	2	0.5	0.0 - 1.4
	Female	0	0.0	
X-linked Ichthyosis	Male	2	0.5	0.0 - 1.4
	Female	0	0.0	
Total Other Cases[5]		14		
Total Cases		903		

[1] Cases can be included in more than one defect category. Cases are counted once within a defect category, therefore system totals will not equal sum of defect categories

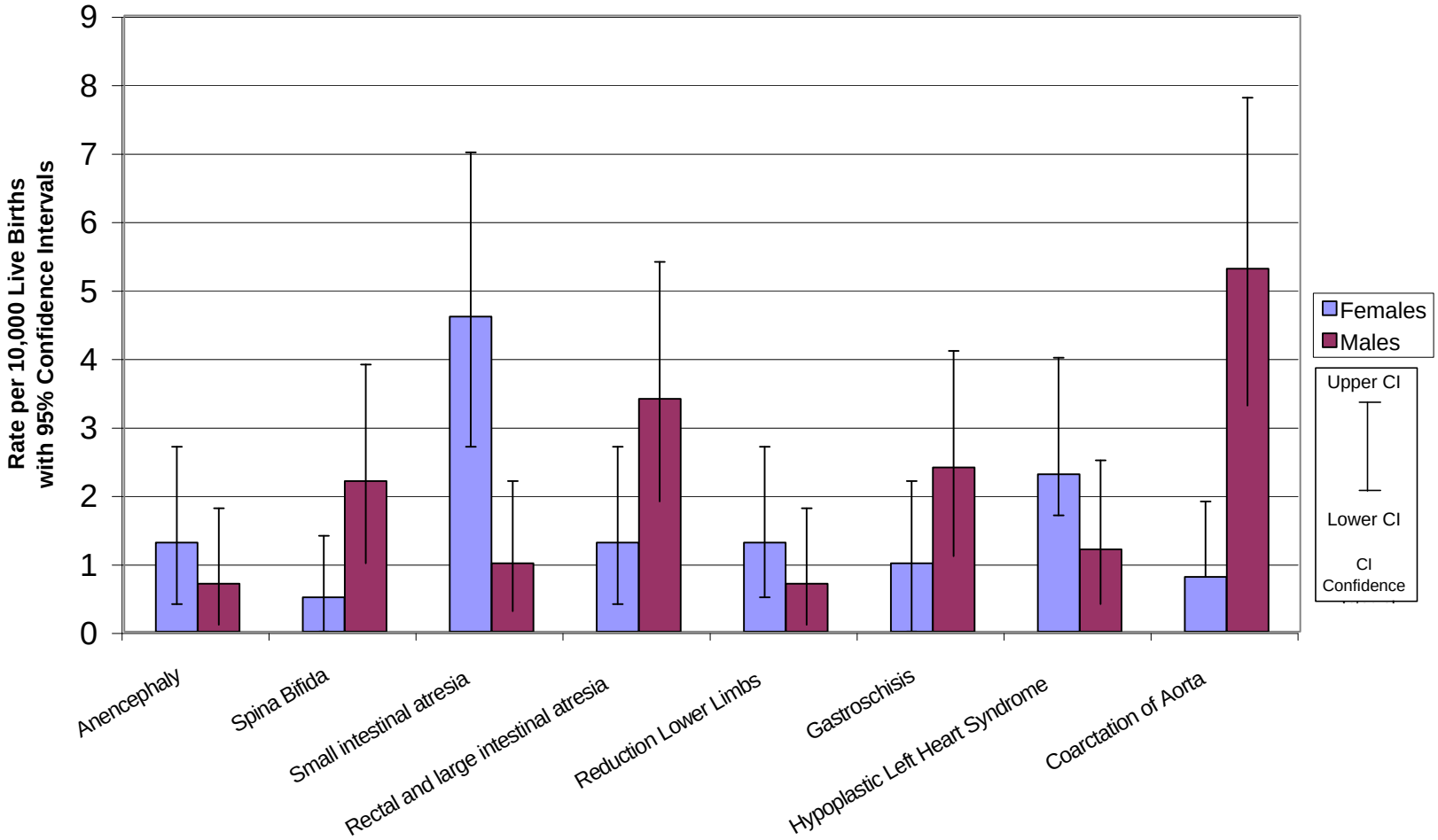
[2] Does not include infants whose sex was unknown (N=1)

[3] Rate per 10,000 live births

[4] Spina bifida includes cases with and without associated hydrocephaly

[5] Total categories in this table include other defects within the system category which have not been selected for presentation. See Appendix for complete list of defects included in these totals.

Figure 4 Selected Birth Defects By Sex of Infant Among Live Births and Stillbirths, Massachusetts: 1999



Chapter 4

Prevalence of Birth Defects by Maternal Age and Race / Ethnicity

Mother's Age

The prevalence of birth defects varies by mother's age. Rates per 10,000 live births are 139.6 for less than 20 years old, 168.5 for 20-24 years old, 129.0 for 25-29 years old, 115.8 for 30-34 years old, and 175.5 for women 35 years and older. Table 8 shows the rates for birth defects by mother's age.

As expected, there is a strong association of Down Syndrome with advanced mother's age (see Figure 5). Women 35 years and older have a live birth Down Syndrome rate of 24.5 per 10,000 births. This rate is five times that of any other maternal age group. Figure 6 shows that younger mothers have a higher share (64%) of gastroschisis cases. This association has been shown in previous studies (Forrester 1997). While results for other defects also differed by age group, the small numbers from one year of surveillance are not sufficient for interpretation.

Table 9 portrays the more common birth defects by mother's age groups. All maternal age groups share common defects: atrial septal defects, venticular septal defects, and cleft lip and cleft palate. Mothers younger than 25 years of age have an aortic valve stenosis rate seven times higher than other age groups. However, one must be cautious when interpreting such small numbers.

Examining birth defects by maternal age is important to monitor since the number of births to older mothers is increasing over time in Massachusetts.

Mother's Race / Hispanic Ethnicity

Table 10 shows the variation in prevalence of birth defects by mother's race and Hispanic ethnicity. The rate per 10,000 live births is 122.5 for Hispanics, 107.8 for Blacks, 105.3 for Whites and 91.8 for Asians. Table 11 shows the more common defects by mother's race and Hispanic ethnicity. More common defects in Hispanics include septal defects, microcephaly, obstructive genitourinary defects, transposition of great arteries, Down syndrome, and cleft lip. In Blacks, the more common defects include septal defects, Down syndrome, hypospadias, microcephaly, coarctation of aorta, and obstructive genitourinary defects. The more common defects in Whites include septal heart defects, Down syndrome, cleft lip, and cleft palate. In Asians, the more common defects include cleft lip, hypospadias, gastroschisis, and omphalocele.

Table 12 shows the prevalence rates for detailed ethnicity groups. Please note that information is collected on discrete ethnic groups as identified on the birth certificate. Overall, birth defects prevalence also varies by ethnicities other than Black, White, Asian, and Hispanic; however, due to the small numbers for specific defects, more data are required on other racial or ethnic groups.

Several factors may contribute to differences in racial and ethnic groups including genetic variation, diet and lifestyle differences, differential access or use of health

care services, or socioeconomic differences. More in-depth studies are needed to understand racial and ethnic patterns.

Table 8 (cont.) Prevalence of Birth Defects by Mother's Age for Live Births, Massachusetts: 1999

Defect[1]	Mother's Age (Years)[2]	Cases	Rate[3]	95% Confidence Interval
CENTRAL NERVOUS SYSTEM				
Anencephaly	<20	1	1.8	0.0 - 7.0
	20-24	1	0.8	0.0 - 3.3
	25-29	3	1.5	0.3 - 3.6
	30-34	1	0.4	0.0 - 1.5
	35+	1	0.6	0.0 - 2.3
Spina Bifida	<20	1	1.8	0.0 - 7.0
	20-24	1	0.8	0.0 - 3.3
	25-29	5	2.5	0.8 - 5.1
	30-34	1	0.4	0.0 - 1.5
	35+	3	1.8	0.3 - 4.4
Hydrocephaly [4]	<20	1	1.8	0.0 - 7.0
	20-24	3	2.5	0.5 - 6.2
	25-29	6	2.9	1.1 - 5.8
	30-34	6	2.3	0.8 - 4.5
	35+	3	1.8	0.3 - 4.4
Encephalocele	<20	0	0.0	
	20-24	0	0.0	
	25-29	1	0.5	0.0 - 1.9
	30-34	3	1.1	0.2 - 2.8
	35+	0	0.0	
Microcephaly	<20	1	1.8	0.0 - 7.0
	20-24	8	6.8	2.9 - 12.3
	25-29	2	1.0	0.1 - 2.8
	30-34	6	2.3	0.8 - 4.5
	35+	2	1.2	0.1 - 3.4
Holoprosencephaly	<20	0	0.0	
	20-24	2	1.7	0.2 - 4.9
	25-29	1	0.5	0.0 - 1.9
	30-34	3	1.1	0.2 - 2.8
	35+	0	0.0	
Tethered Cord	<20	0	0.0	
	20-24	0	0.0	
	25-29	3	1.5	0.3 - 3.6
	30-34	2	0.8	0.1 - 2.2
	35+	0	0.0	
Total Central Nervous System Cases[5]		89		
EYE				
Anophthalmia/Microphthalmia	<20	0	0.0	
	20-24	0	0.0	
	25-29	1	0.5	0.0 - 1.9
	30-34	3	1.1	0.2 - 2.8
	35+	0	0.0	
Congenital Cataract	<20	1	1.8	0.0 - 7.0
	20-24	3	2.5	0.5 - 6.2
	25-29	1	0.5	0.0 - 1.9
	30-34	4	1.5	0.4 - 3.4
	35+	2	1.2	0.1 - 3.4

Table 8 (cont.) Prevalence of Birth Defects by Mother's Age for Live Births, Massachusetts: 1999

Defect[1]	Mother's Age (Years)[2]	Cases	Rate[3]	95% Confidence Interval
Aniridia	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	0	0.0	
	30-34	0	0.0	
	35+	0	0.0	
Coloboma	<20	0	0.0	
	20-24	0	0.0	
	25-29	0	0.0	
	30-34	3	1.1	0.2 - 2.8
	35+	3	1.8	0.3 - 4.4
Total Eye Cases[5]		24		
EAR				
Anotia/Microtia	<20	0	0.0	
	20-24	2	1.7	0.2 - 4.9
	25-29	2	1.0	0.1 - 2.8
	30-34	4	1.5	0.4 - 3.4
	35+	3	1.8	0.3 - 4.4
Total Ear Cases[5]		13		
CARDIOVASCULAR/RESPIRATORY				
Common Truncus/Aortic Septal Defects	<20	0	0.0	
	20-24	0	0.0	
	25-29	0	0.0	
	30-34	1	0.4	0.0 - 1.5
	35+	0	0.0	
Transposition of Great Arteries	<20	2	3.6	0.3 - 10.3
	20-24	3	2.5	0.5 - 6.2
	25-29	7	3.4	1.4 - 6.4
	30-34	5	1.9	0.6 - 3.9
	35+	4	2.4	0.6 - 5.3
Tetralogy of Fallot	<20	1	1.8	0.0 - 7.0
	20-24	0	0.0	
	25-29	7	3.4	1.4 - 6.4
	30-34	8	3.0	1.3 - 5.5
	35+	8	4.8	2.0 - 8.7
Ventricular Septal Defect	<20	5	8.9	2.8 - 18.5
	20-24	10	8.5	4.0 - 14.5
	25-29	13	6.4	3.4 - 10.3
	30-34	19	7.2	4.3 - 10.8
	35+	21	12.5	7.7 - 18.5
Atrial Septal Defect	<20	7	12.5	5.0 - 23.5
	20-24	16	13.5	7.7 - 21.0
	25-29	22	10.8	6.8 - 15.8
	30-34	14	5.3	2.9 - 8.5
	35+	32	19.1	13.1 - 26.3
Endocardial Cushion Defect	<20	1	1.8	0.0 - 7.0
	20-24	2	1.7	0.2 - 4.9
	25-29	5	2.5	0.8 - 5.1
	30-34	6	2.3	0.8 - 4.5
	35+	10	6.0	2.8 - 10.2

Table 8 (cont.) Prevalence of Birth Defects by Mother's Age for Live Births, Massachusetts: 1999

Defect[1]	Mother's Age (Years)[2]	Cases	Rate[3]	95% Confidence Interval
Pulmonary Valve Atresia and Stenosis	<20	2	3.6	0.3 - 10.3
	20-24	7	5.9	2.3 - 11.1
	25-29	10	4.9	2.3 - 8.4
	30-34	6	2.3	0.8 - 4.5
	35+	8	4.8	2.0 - 8.7
Tricuspid Valve Atresia and Stenosis	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	1	0.5	0.0 - 1.9
	30-34	3	1.1	0.2 - 2.8
	35+	2	1.2	0.1 - 3.4
Ebstein Anomaly	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	1	0.5	0.0 - 1.9
	30-34	1	0.4	0.0 - 1.5
	35+	1	0.6	0.0 - 2.3
Aortic Valve Stenosis	<20	4	7.2	1.9 - 15.9
	20-24	1	0.8	0.0 - 3.3
	25-29	2	1.0	0.1 - 2.8
	30-34	3	1.1	0.2 - 2.8
	35+	2	1.2	0.1 - 3.4
Hypoplastic Left Heart Syndrome	<20	2	3.6	0.3 - 10.3
	20-24	2	1.7	0.2 - 4.9
	25-29	3	1.5	0.3 - 3.6
	30-34	2	0.8	0.1 - 2.2
	35+	3	1.8	0.3 - 4.4
Patent Ductus Arteriosus	<20	6	10.7	3.9 - 21.0
	20-24	17	14.4	8.4 - 22.0
	25-29	25	12.3	7.9 - 17.5
	30-34	27	10.3	6.8 - 14.5
	35+	28	16.7	11.1 - 23.5
Coarctation of Aorta	<20	1	1.8	0.0 - 7.0
	20-24	6	5.1	1.8 - 10.0
	25-29	4	2.0	0.5 - 4.4
	30-34	10	3.8	1.8 - 6.5
	35+	4	2.4	0.6 - 5.3
Total Anomalous Pulmonary Venous Return	<20	0	0.0	
	20-24	0	0.0	
	25-29	1	0.5	0.0 - 1.9
	30-34	1	0.4	0.0 - 1.5
	35+	3	1.8	0.3 - 4.4
Partial Anomalous Pulmonary Venous Return	<20	0	0.0	
	20-24	0	0.0	
	25-29	3	1.5	0.3 - 3.6
	30-34	0	0.0	
	35+	1	0.6	0.0 - 2.3
Single Ventricle	<20	0	0.0	
	20-24	0	0.0	
	25-29	2	1.0	0.1 - 2.8
	30-34	1	0.4	0.0 - 1.5
	35+	0	0.0	

Table 8 (cont.) Prevalence of Birth Defects by Mother's Age for Live Births, Massachusetts: 1999

Defect[1]	Mother's Age (Years)[2]	Cases	Rate[3]	95% Confidence Interval
Situs Inversus/Heterotaxia	<20	0	0.0	
	20-24	2	1.7	0.2 - 4.9
	25-29	3	1.5	0.3 - 3.6
	30-34	3	1.1	0.2 - 2.8
	35+	3	1.8	0.3 - 4.4
Total Cardiovascular/ Respiratory Cases[5]		300		
OROFACIAL				
Cleft Palate without Cleft Lip	<20	3	5.4	1.0 - 13.2
	20-24	7	5.9	2.3 - 11.1
	25-29	11	5.4	2.7 - 9.1
	30-34	14	5.3	2.9 - 8.5
	35+	9	5.4	2.4 - 9.5
Cleft Lip with and without Cleft Palate	<20	6	10.7	3.9 - 21.0
	20-24	13	11.0	5.8 - 17.8
	25-29	14	6.9	3.7 - 10.9
	30-34	18	6.8	4.0 - 10.4
	35+	16	9.6	5.4 - 14.8
Choanal Atresia	<20	0	0.0	
	20-24	0	0.0	
	25-29	0	0.0	
	30-34	2	0.8	0.1 - 2.2
	35+	0	0.0	
Pierre Robin Sequence	<20	0	0.0	
	20-24	2	1.7	0.2 - 4.9
	25-29	4	2.0	0.5 - 4.4
	30-34	5	1.9	0.6 - 3.9
	35+	4	2.4	0.6 - 5.3
Branchial Cleft/Sinus	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	3	1.5	0.3 - 3.6
	30-34	1	0.4	0.0 - 1.5
	35+	3	1.8	0.3 - 4.4
Total Orofacial Cases[5]		124		
GASTROINTESTINAL				
Esophageal Atresia/Tracheoesophageal Fistula	<20	0	0.0	
	20-24	3	2.5	0.5 - 6.2
	25-29	6	2.9	1.1 - 5.8
	30-34	6	2.3	0.8 - 4.5
	35+	5	3.0	0.9 - 6.2
Rectal and Large Intestinal Atresia/Stenosis	<20	1	1.8	0.0 - 7.0
	20-24	4	3.4	0.9 - 7.5
	25-29	3	1.5	0.3 - 3.6
	30-34	5	1.9	0.6 - 3.9
	35+	4	2.4	0.6 - 5.3
Hirschsprung Disease	<20	2	3.6	0.3 - 10.3
	20-24	3	2.5	0.5 - 6.2
	25-29	4	2.0	0.5 - 4.4
	30-34	0	0.0	
	35+	1	0.6	0.0 - 2.3

Table 8 (cont.) Prevalence of Birth Defects by Mother's Age for Live Births, Massachusetts: 1999

Defect[1]	Mother's Age (Years)[2]	Cases	Rate[3]	95% Confidence Interval
Biliary Atresia	<20	1	1.8	0.0 - 7.0
	20-24	1	0.8	0.0 - 3.3
	25-29	0	0.0	
	30-34	1	0.4	0.0 - 1.5
	35+	0	0.0	
Small Intestinal Atresia	<20	2	3.6	0.3 - 10.3
	20-24	3	2.5	0.5 - 6.2
	25-29	6	2.9	1.1 - 5.8
	30-34	8	3.0	1.3 - 5.5
	35+	2	1.2	0.1 - 3.4
Total Gastrointestinal Cases[5]		87		
GENITOURINARY				
Renal Agenesis/Hypoplasia	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	1	0.5	0.0 - 1.9
	30-34	1	0.4	0.0 - 1.5
	35+	0	0.0	
Bladder Exstrophy	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	0	0.0	
	30-34	0	0.0	
	35+	0	0.0	
Obstructive Genitourinary Defect	<20	7	12.5	5.0 - 23.5
	20-24	3	2.5	0.5 - 6.2
	25-29	11	5.4	2.7 - 9.1
	30-34	10	3.8	1.8 - 6.5
	35+	11	6.6	3.3 - 11.0
Hypospadias	<20	4	7.2	1.9 - 15.9
	20-24	6	5.1	1.8 - 10.0
	25-29	12	5.9	3.0 - 9.7
	30-34	16	6.1	3.5 - 9.4
	35+	10	6.0	2.8 - 10.2
Ambiguous Genitalia	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	0	0.0	
	30-34	2	0.8	0.1 - 2.2
	35+	2	1.2	0.1 - 3.4
Total Genitourinary Cases[5]		116		
MUSCULOSKELETAL				
Reduction Deformity, Upper Limbs	<20	1	1.8	0.0 - 7.0
	20-24	4	3.4	0.9 - 7.5
	25-29	3	1.5	0.3 - 3.6
	30-34	4	1.5	0.4 - 3.4
	35+	2	1.2	0.1 - 3.4
Reduction Deformity, Lower Limbs	<20	0	0.0	
	20-24	2	1.7	0.2 - 4.9
	25-29	2	1.0	0.1 - 2.8
	30-34	1	0.4	0.0 - 1.5
	35+	1	0.6	0.0 - 2.3

Table 8 (cont.) Prevalence of Birth Defects by Mother's Age for Live Births, Massachusetts: 1999

Defect[1]	Mother's Age (Years)[2]	Cases	Rate[3]	95% Confidence Interval
Gastroschisis	<20	2	3.6	0.3 - 10.3
	20-24	7	5.9	2.3 - 11.1
	25-29	4	2.0	0.5 - 4.4
	30-34	1	0.4	0.0 - 1.5
	35+	0	0.0	
Omphalocele	<20	0	0.0	
	20-24	3	2.5	0.5 - 6.2
	25-29	3	1.5	0.3 - 3.6
	30-34	2	0.8	0.1 - 2.2
	35+	3	1.8	0.3 - 4.4
Diaphragmatic Hernia	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	5	2.5	0.8 - 5.1
	30-34	7	2.7	1.1 - 5.0
	35+	3	1.8	0.3 - 4.4
Achondroplasia	<20	0	0.0	
	20-24	0	0.0	
	25-29	1	0.5	0.0 - 1.9
	30-34	0	0.0	
	35+	1	0.6	0.0 - 2.3
Thanatophoric Dysplasia	<20	0	0.0	
	20-24	0	0.0	
	25-29	0	0.0	
	30-34	1	0.4	0.0 - 1.5
	35+	0	0.0	
Osteogenesis Imperfecta	<20	0	0.0	
	20-24	0	0.0	
	25-29	1	0.5	0.0 - 1.9
	30-34	1	0.4	0.0 - 1.5
	35+	0	0.0	
Craniosynostosis	<20	2	3.6	0.3 - 10.3
	20-24	2	1.7	0.2 - 4.9
	25-29	2	1.0	0.1 - 2.8
	30-34	7	2.7	1.1 - 5.0
	35+	1	0.6	0.0 - 2.3
Total Musculoskeletal Cases[5]		227		
CHROMOSOMAL/SYNDROMES				
Trisomy 13	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	0	0.0	
	30-34	0	0.0	
	35+	1	0.6	0.0 - 2.3
Trisomy 21 (Down Syndrome)	<20	2	3.6	0.3 - 10.3
	20-24	3	2.5	0.5 - 6.2
	25-29	6	2.9	1.1 - 5.8
	30-34	13	4.9	2.6 - 8.0
	35+	41	24.5	17.6 - 32.5

Table 8 (cont.) Prevalence of Birth Defects by Mother's Age for Live Births, Massachusetts: 1999

Defect[1]	Mother's Age (Years)[2]	Cases	Rate[3]	95% Confidence Interval
Trisomy 18	<20	1	1.8	0.0 - 7.0
	20-24	1	0.8	0.0 - 3.3
	25-29	2	1.0	0.1 - 2.8
	30-34	1	0.4	0.0 - 1.5
	35+	6	3.6	1.3 - 7.0
Turner Syndrome	<20	1	1.8	0.0 - 7.0
	20-24	2	1.7	0.2 - 4.9
	25-29	0	0.0	
	30-34	2	0.8	0.1 - 2.2
	35+	2	1.2	0.1 - 3.4
Klinefelter Syndrome	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	1	0.5	0.0 - 1.9
	30-34	0	0.0	
	35+	7	4.2	1.7 - 7.8
DiGeorge Syndrome	<20	0	0.0	
	20-24	0	0.0	
	25-29	4	2.0	0.5 - 4.4
	30-34	1	0.4	0.0 - 1.5
	35+	1	0.6	0.0 - 2.3
Total Chromosomal/Syndromes Cases[5]		145		
OTHER				
Amniotic Bands	<20	0	0.0	
	20-24	3	2.5	0.5 - 6.2
	25-29	0	0.0	
	30-34	1	0.4	0.0 - 1.5
	35+	0	0.0	
Endocrine Anomalies	<20	0	0.0	
	20-24	1	0.8	0.0 - 3.3
	25-29	0	0.0	
	30-34	1	0.4	0.0 - 1.5
	35+	1	0.6	0.0 - 2.3
Epidermolysis Bullosa	<20	0	0.0	
	20-24	0	0.0	
	25-29	1	0.5	0.0 - 1.9
	30-34	0	0.0	
	35+	1	0.6	0.0 - 2.3
X-linked Ichthyosis	<20	0	0.0	
	20-24	0	0.0	
	25-29	1	0.5	0.0 - 1.9
	30-34	1	0.4	0.0 - 1.5
	35+	0	0.0	
Total Other Cases[5]		11		

[1] Cases can be included in more than one defect category. Cases are counted once within a defect category, therefore system totals will not equal sum of defect categories

[2] Three mothers <15 years of age included in <20 age category; four mothers > 44 years of age included in 35+ category

[3] Rate per 10,000 live births

[4] Spina bifida includes cases with and without associated hydrocephaly

[5] Total categories in this table include other defects within the system category which have not been selected for presentation. See Appendix for complete list of defects included in these totals

Table 9 Most Common Defects by Mother's Age, Massachusetts: 1999[1]

Mother's Age	Defect	Count	Rate[2]	95% Confidence Interval
<20 years				
	Atrial Septal Defect	7	12.5	5.0 - 23.5
	Obstructive Genitourinary Defect	7	12.5	5.0 - 23.5
	Cleft Lip with and without Cleft Palate	6	10.7	3.9 - 21.0
	Ventricular Septal Defect	5	8.9	2.8 - 18.5
	Aortic Valve Stenosis	4	7.2	1.9 - 15.9
	Hypospadias (2nd and 3rd Degree)	4	7.2	1.9 - 15.9
20-24 years				
	Atrial Septal Defect	16	13.5	7.7 - 21.0
	Cleft Lip with and without Cleft Palate	13	11.0	5.8 - 17.8
	Ventricular Septal Defect	10	8.5	4.0 - 14.5
	Microcephaly	8	6.8	2.9 - 12.3
	Gastroschisis	7	5.9	2.3 - 11.1
	Pulmonary Valve Atresia and Stenosis	7	5.9	2.3 - 11.1
	Cleft Palate without Cleft Lip	7	5.9	2.3 - 11.1
25-29 years				
	Atrial Septal Defect	22	10.8	6.8 - 15.8
	Cleft Lip with and without Cleft Palate	14	6.9	3.7 - 10.9
	Ventricular Septal Defect	13	6.4	3.4 - 10.3
	Hypospadias (2nd and 3rd Degree)	12	5.9	3.0 - 9.7
	Cleft Palate without Cleft Lip	11	5.4	2.7 - 9.1
	Obstructive Genitourinary Defect	11	5.4	2.7 - 9.1
30-34 years				
	Ventricular Septal Defect	19	7.2	4.3 - 10.8
	Cleft Lip with and without Cleft Palate	18	6.8	4.0 - 10.4
	Hypospadias (2nd and 3rd Degree)	16	6.1	3.5 - 9.4
	Atrial Septal Defect	14	5.3	2.9 - 8.5
	Cleft Palate without Cleft Lip	14	5.3	2.9 - 8.5
35+ years				
	Trisomy 21 (Down Syndrome)	41	24.5	17.6 - 32.5
	Atrial Septal Defect	32	19.1	13.1 - 26.3
	Ventricular Septal Defect	21	12.5	7.7 - 18.5
	Cleft Lip with and without Cleft Palate	16	9.6	5.4 - 14.8
	Obstructive Gentiourinary	11	6.6	3.3 - 11.0

[1] Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

[2] Rate per 10,000 live births

Figure 5 Distribution of Down Syndrome and Distribution of Births Among Mother's Age Groups, Massachusetts: 1999

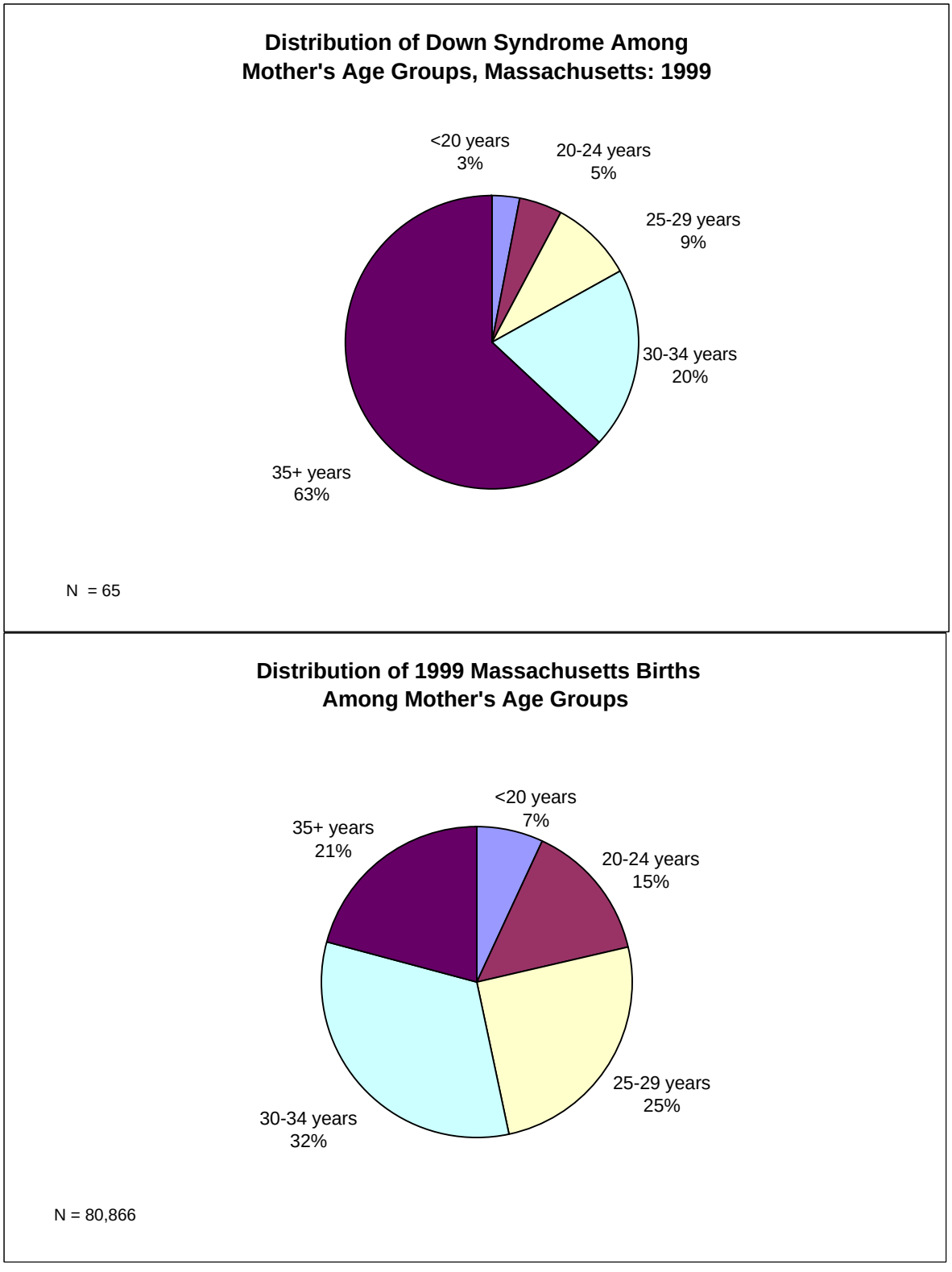


Figure 6 Distribution of Gastroschisis and Distribution of Births By Mother's Age, Massachusetts: 1999

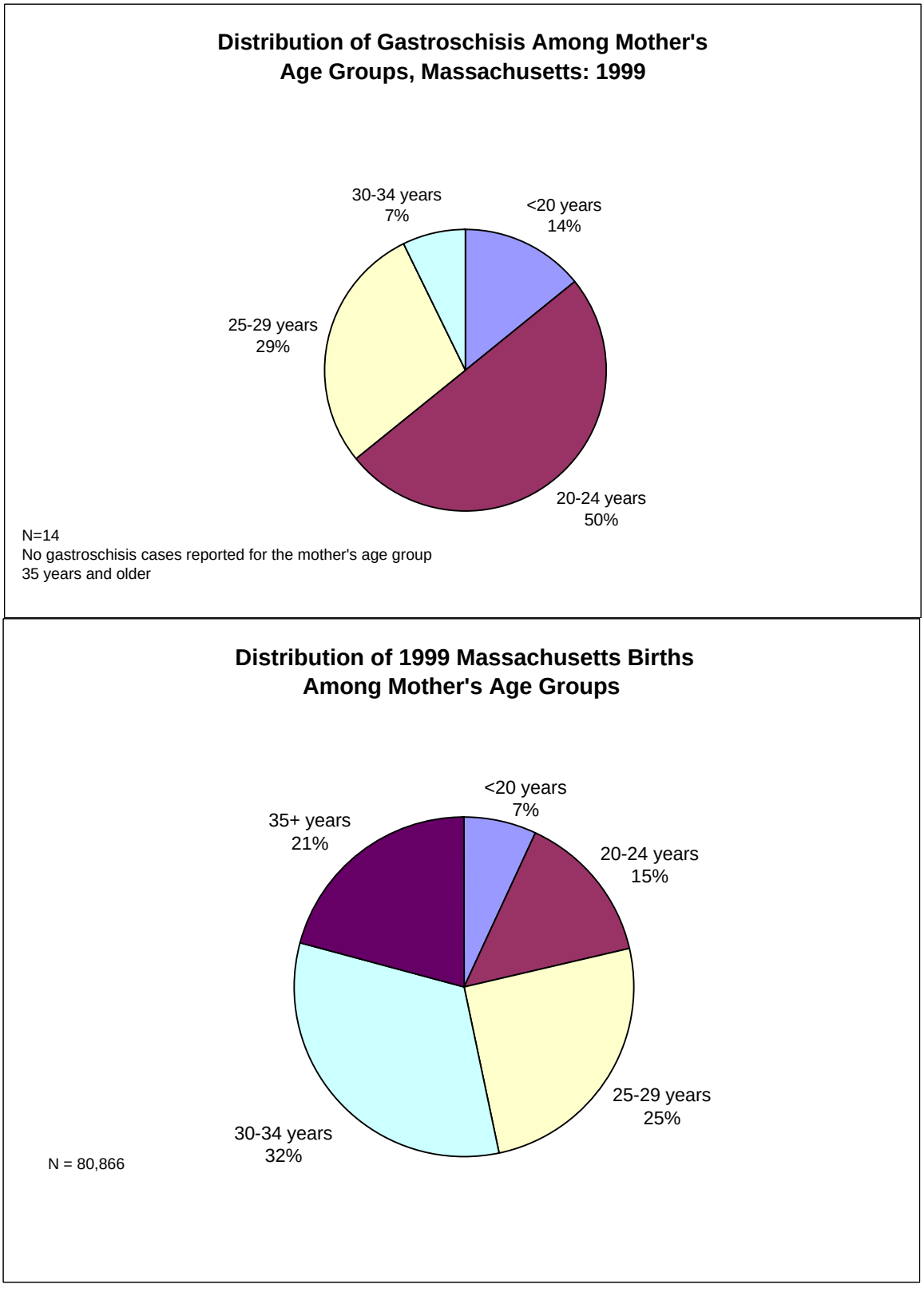


Table 10 Prevalence of Birth Defects by Mother's Race/Hispanic Ethnicity for Livebirths, Massachusetts: 1999

Defect[1]	Mother's Race/Ethnicity[2,3]	Cases	Rate[4]	95% Confidence Interval
CENTRAL NERVOUS SYSTEM				
Anencephaly	White	4	0.7	0.2 - 1.5
	Black	2	3.4	0.3 - 9.8
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Spina Bifida	White	10	1.7	0.8 - 2.8
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Hydrocephaly[5]	White	15	2.5	1.4 - 3.9
	Black	1	1.7	0.0 - 6.7
	Hispanic	2	2.3	0.2 - 6.5
	Asian	1	2.4	0.0 - 9.5
Encephalocele	White	4	0.7	0.2 - 1.5
	Black	0	0.0	
	Hispanic	0	0.0	
	Asian	0	0.0	
Microcephaly	White	11	1.8	0.9 - 3.1
	Black	3	5.1	1.0 - 12.6
	Hispanic	5	5.7	1.8 - 11.7
	Asian	0	0.0	
Holoprosencephaly	White	4	0.7	0.2 - 1.5
	Black	1	1.7	0.0 - 6.7
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Tethered Cord	White	4	0.7	0.2 - 1.5
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Total Central Nervous System Cases[6]		89		
EYE				
Anophthalmia/Microphthalmia	White	3	0.5	0.1 - 1.2
	Black	0	0.0	
	Hispanic	0	0.0	
	Asian	1	2.4	0.0 - 9.5
Congenital Cataract	White	9	1.5	0.7 - 2.6
	Black	1	1.7	0.0 - 6.7
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Aniridia	White	1	0.2	0.0 - 0.6
	Black	0	0.0	
	Hispanic	0	0.0	
	Asian	0	0.0	

Table 10 Prevalence of Birth Defects by Mother's Race/Hispanic Ethnicity for Livebirths, Massachusetts: 1999

Defect[1]	Mother's Race/Ethnicity[2,3]	Cases	Rate[4]	95% Confidence Interval
Coloboma	White	5	0.8	0.3 - 1.7
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Total Eye Cases[6]		24		
EAR				
Anotia/Microtia	White	7	1.2	0.5 - 2.2
	Black	0	0.0	
	Hispanic	3	3.4	0.6 - 8.3
	Asian	1	2.4	0.0 - 9.5
Total Ear Cases[6]		13		
CARDIOVASCULAR/RESPIRATORY				
Common Truncus/Aortic Septal Defects	White	1	0.2	0.0 - 0.6
	Black	0	0.0	
	Hispanic	0	0.0	
	Asian	0	0.0	
Transposition of Great Arteries	White	16	2.6	1.5 - 4.1
	Black	0	0.0	
	Hispanic	5	5.7	1.8 - 11.7
	Asian	0	0.0	
Tetralogy of Fallot	White	21	3.5	2.1 - 5.1
	Black	1	1.7	0.0 - 6.7
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Ventricular Septal Defect	White	50	8.3	6.1 - 10.7
	Black	6	10.3	3.7 - 20.1
	Hispanic	10	11.3	5.4 - 19.5
	Asian	0	0.0	
Atrial Septal Defect	White	69	11.4	8.9 - 14.3
	Black	4	6.8	1.8 - 15.2
	Hispanic	15	17.0	9.5 - 26.7
	Asian	1	2.4	0.0 - 9.5
Endocardial Cushion Defect	White	20	3.3	2.0 - 4.9
	Black	1	1.7	0.0 - 6.7
	Hispanic	2	2.3	0.2 - 6.5
	Asian	1	2.4	0.0 - 9.5
Pulmonary Valve Atresia and Stenosis	White	24	4.0	2.5 - 5.7
	Black	2	3.4	0.3 - 9.8
	Hispanic	4	4.5	1.2 - 10.1
	Asian	1	2.4	0.0 - 9.5
Tricuspid Valve Atresia and Stenosis	White	5	0.8	0.3 - 1.7
	Black	0	0.0	
	Hispanic	2	2.3	0.2 - 6.5
	Asian	0	0.0	
Ebstein Anomaly	White	3	0.5	0.1 - 1.2
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	

Table 10 Prevalence of Birth Defects by Mother's Race/Hispanic Ethnicity for Livebirths, Massachusetts: 1999

Defect[1]	Mother's Race/Ethnicity[2,3]	Cases	Rate[4]	95% Confidence Interval
Aortic Valve Stenosis	White	10	1.7	0.8 - 2.8
	Black	1	1.7	0.0 - 6.7
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Hypoplastic Left Heart Syndrome	White	9	1.5	0.7 - 2.6
	Black	2	3.4	0.3 - 9.8
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Patent Ductus Arteriosus	White	75	12.4	9.8 - 15.4
	Black	6	10.3	3.7 - 20.1
	Hispanic	15	17.0	9.5 - 26.7
	Asian	4	9.7	2.5 - 21.5
Coarctation of Aorta	White	20	3.3	2.0 - 4.9
	Black	3	5.1	1.0 - 12.6
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Total Anomalous Pulmonary Venous Return	White	5	0.8	0.3 - 1.7
	Black	0	0.0	
	Hispanic	0	0.0	
	Asian	0	0.0	
Partial Anomalous Pulmonary Venous Return	White	2	0.3	0.0 - 0.9
	Black	1	1.7	0.0 - 6.7
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Single Ventricle	White	2	0.3	0.0 - 0.9
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Situs Inversus/Heterotaxia	White	5	0.8	0.3 - 1.7
	Black	2	3.4	0.3 - 9.8
	Hispanic	2	2.3	0.2 - 6.5
	Asian	1	2.4	0.0 - 9.5
Total Cardiovascular/Respiratory Cases[6]		300		
OROFACIAL				
Cleft Palate without Cleft Lip	White	38	6.3	4.4 - 8.5
	Black	1	1.7	0.0 - 6.7
	Hispanic	3	3.4	0.6 - 8.3
	Asian	1	2.4	0.0 - 9.5
Cleft Lip with and without Cleft Palate	White	48	7.9	5.9 - 10.4
	Black	1	1.7	0.0 - 6.7
	Hispanic	11	12.5	6.2 - 20.9
	Asian	5	12.1	3.8 - 25.0
Choanal Atresia	White	2	0.3	0.0 - 0.9
	Black	0	0.0	
	Hispanic	0	0.0	
	Asian	0	0.0	

Table 10 Prevalence of Birth Defects by Mother's Race/Hispanic Ethnicity for Livebirths, Massachusetts: 1999

Defect[1]	Mother's Race/Ethnicity[2,3]	Cases	Rate[4]	95% Confidence Interval
Pierre Robin Sequence	White	14	2.3	1.3 - 3.7
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Brachial Cleft/Sinus	White	6	1.0	0.4 - 1.9
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	1	2.4	0.0 - 9.5
Total Orofacial Cases[6]		124		
GASTROINTESTINAL				
Esophageal Atresia/Tracheoesophageal Fistula	White	16	2.6	1.5 - 4.1
	Black	1	1.7	0.0 - 6.7
	Hispanic	2	2.3	0.2 - 6.5
	Asian	1	2.4	0.0 - 9.5
Rectal and Large Intestinal Atresia/Stenosis	White	12	2.0	1.0 - 3.3
	Black	0	0.0	
	Hispanic	3	3.4	0.6 - 8.3
	Asian	1	2.4	0.0 - 9.5
Hirschsprung Disease	White	4	0.7	0.2 - 1.5
	Black	2	3.4	0.3 - 9.8
	Hispanic	2	2.3	0.2 - 6.5
	Asian	0	0.0	
Biliary Atresia	White	1	0.2	0.0 - 0.6
	Black	1	1.7	0.0 - 6.7
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Small Intestinal Atresia	White	17	2.8	1.6 - 4.3
	Black	0	0.0	
	Hispanic	3	3.4	0.6 - 8.3
	Asian	1	2.4	0.0 - 9.5
Total Gastrointestinal System Cases[6]		87		
GENITOURINARY				
Renal Agenesis/Hypoplasia	White	3	0.5	0.1 - 1.2
	Black	0	0.0	
	Hispanic	0	0.0	
	Asian	0	0.0	
Bladder Exstrophy	White	0	0.0	
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Obstructive Genitourinary Defect	White	29	4.8	3.2 - 6.7
	Black	3	5.1	1.0 - 12.6
	Hispanic	6	6.8	2.4 - 13.3
	Asian	1	2.4	0.0 - 9.5
Hypospadias	White	34	5.6	3.9 - 7.7
	Black	4	6.8	1.8 - 15.2
	Hispanic	4	4.5	1.2 - 10.1
	Asian	5	12.1	3.8 - 25.0

Table 10 Prevalence of Birth Defects by Mother's Race/Hispanic Ethnicity for Livebirths, Massachusetts: 1999

Defect[1]	Mother's Race/Ethnicity[2,3]	Cases	Rate[4]	95% Confidence Interval
Ambiguous Genitalia	White	2	0.3	0.0 - 0.9
	Black	0	0.0	
	Hispanic	3	3.4	0.6 - 8.3
	Asian	0	0.0	
Total Genitourinary System Cases[6]		116		
MUSCULOSKELETAL				
Reduction Deformity, Upper Limbs	White	10	1.7	0.8 - 2.8
	Black	1	1.7	0.0 - 6.7
	Hispanic	2	2.3	0.2 - 6.5
	Asian	1	2.4	0.0 - 9.5
Reduction Deformity, Lower Limbs	White	3	0.5	0.1 - 1.2
	Black	2	3.4	0.3 - 9.8
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Gastroschisis	White	11	1.8	0.9 - 3.1
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	2	4.8	0.5 - 13.9
Omphalocele	White	6	1.0	0.4 - 1.9
	Black	1	1.7	0.0 - 6.7
	Hispanic	1	1.1	0.0 - 4.4
	Asian	2	4.8	0.5 - 13.9
Diaphragmatic Hernia	White	14	2.3	1.3 - 3.7
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	1	2.4	0.0 - 9.5
Achondroplasia	White	0	0.0	
	Black	1	1.7	0.0 - 6.7
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Thanatophoric Dysplasia	White	0	0.0	
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Osteogenesis Imperfecta	White	1	0.2	0.0 - 0.6
	Black	0	0.0	0.0 - 1.6
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Craniosynostosis	White	12	2.0	1.0 - 3.3
	Black	1	1.7	0.0 - 6.7
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Total Musculoskeletal System Cases[6]		227		
CHROMOSOMAL/SYNDROMES				
Trisomy 13	White	1	0.2	0.0 - 0.6
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	

Table 10 Prevalence of Birth Defects by Mother's Race/Hispanic Ethnicity for Livebirths, Massachusetts: 1999

Defect[1]	Mother's Race/Ethnicity[2,3]	Cases	Rate[4]	95% Confidence Interval
Trisomy 21 (Down Syndrome)	White	51	8.4	6.3 - 10.9
	Black	5	8.6	2.7 - 17.7
	Hispanic	5	5.7	1.8 - 11.7
	Asian	1	2.4	0.0 - 9.5
Trisomy 18	White	8	1.3	0.6 - 2.4
	Black	0	0.0	
	Hispanic	2	2.3	0.2 - 6.5
	Asian	0	0.0	
Turner Syndrome	White	5	0.8	0.3 - 1.7
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	1	2.4	0.0 - 9.5
Klinefelter Syndrome	White	9	1.5	0.7 - 2.6
	Black	0	0.0	
	Hispanic	0	0.0	
	Asian	0	0.0	
DiGeorge Syndrome	White	5	0.8	0.3 - 1.7
	Black	1	1.7	0.0 - 6.7
	Hispanic	0	0.0	
	Asian	0	0.0	
Total Chromosomal/Syndromes Cases[6]		145		
OTHER				
Amniotic Bands	White	4	0.7	0.2 - 1.5
	Black	0	0.0	
	Hispanic	0	0.0	
	Asian	0	0.0	
Endocrine Anomalies	White	2	0.3	0.0 - 0.9
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	0	0.0	
Epidermolysis Bullosa	White	2	0.3	0.0 - 0.9
	Black	0	0.0	
	Hispanic	0	0.0	
	Asian	0	0.0	
X-linked Ichthyosis	White	0	0.0	
	Black	0	0.0	
	Hispanic	1	1.1	0.0 - 4.4
	Asian	1	2.4	0.0 - 9.5
Total Other Cases[6]		11		

[1] Cases can be included in more than one defect category. Cases are counted once within a defect category, therefore system totals will not equal sum of defect categories

[2] White, Black and Asian groups do not include mothers of Hispanic origin

[3] Due to small numbers races classified as "other" are excluded

[4] Rate per 10,000 live births

[5] Spina bifida includes cases with and without associated hydrocephaly

[6] Total categories in this table include other defects within the system category which have not been selected for presentation. See Appendix for complete list of defects included in these totals

Table 11 Most Common Defects by Race/Hispanic Ethnicity[1] Among Live Births, Massachusetts: 1999

Race[2]	Defect	Count	Rate[3]	95 % Confidence Intervals
White, Non-Hispanic				
	Atrial Septal Defect	69	11.4	8.9 - 14.3
	Trisomy 21 (Down Syndrome)	51	8.4	6.3 - 10.9
	Ventricular Septal Defect	50	8.3	6.1 - 10.7
	Cleft Lip with and without Cleft Palate	48	7.9	5.9 - 10.4
	Cleft Palate without Cleft Lip	38	6.3	4.4 - 8.5
Black, Non-Hispanic				
	Ventricular Septal Defect	6	10.3	3.7 - 20.1
	Trisomy 21 (Down Syndrome)	5	8.6	2.7 - 17.7
	Atrial Septal Defect	4	6.8	1.8 - 15.2
	Hypospadias (2nd and 3rd Degree)	4	6.8	1.8 - 15.2
	Microcephaly	3	5.1	1.0 - 12.6
	Coarctation of the Aorta	3	5.1	1.0 - 12.6
	Obstructive Gentourinary	3	5.1	1.0 - 12.6
Hispanic				
	Atrial Septal Defect	15	17.0	9.5 - 26.7
	Cleft Lip with and without Cleft Palate	11	12.5	6.2 - 20.9
	Ventricular Septal Defect	10	11.3	5.4 - 19.5
	Obstructive Genitourinary Defect	6	6.8	2.4 - 13.3
	Microcephaly	5	5.7	1.8 - 11.7
	Transposition of Great Arteries	5	5.7	1.8 - 11.7
	Trisomy 21 (Down Syndrome)	5	5.7	1.8 - 11.7
Asian				
	Cleft Lip with and without Cleft Palate	5	12.1	3.8 - 25.0
	Hypospadias	5	12.1	3.8 - 25.0
	Gastroschisis	2	4.8	0.5 - 13.9
	Omphalocele	2	4.8	0.5 - 13.9

[1] Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

[2] Due to small numbers races classified as "other" are excluded

[3] Rate per 10,000 live births

Table 12 Overall Distribution of Infants with Birth Defects By Mother's Ethnicity, Massachusetts: 1999

Ethnicity	Infants	Rate[1]	95% Confidence Interval
Puerto Rican	55	124.5	93.8 - 159.6
Dominican	16	102.9	58.7 - 159.5
Salvadoran	10	152.0	72.4 - 260.8
Other Central American	9	125.2	56.8 - 220.3
Other Hispanic[2]	18	122.9	72.7 - 186.2
Chinese	13	110.3	58.5 - 178.3
Vietnamese	4	58.6	15.2 - 130.0
Cambodian	7	125.7	49.8 - 236.0
Asian Indian	6	83.7	30.1 - 164.0
Other Asian/Pacific Islander[3]	10	91.6	43.6 - 157.1
Cape Verdean	10	127.7	60.8 - 219.1
Brazilian	9	112.6	51.1 - 198.3
Other Portuguese	7	51.7	20.5 - 97.2
Haitian	13	127.8	67.8 - 206.8
West Indian/Caribbean[4]	6	90.5	32.6 - 177.4
African American	36	124.8	87.3 - 168.9
African[5]	9	109.8	49.8 - 193.2
Middle Easterner[6]	13	164.6	87.3 - 266.2
Native American	5	190.8	60.2 - 394.8
European	154	105.9	89.8 - 123.3

[1] Rate per 10,000 live births

[2] Other Hispanic includes Mexican, Cuban, Colombian and Other South American

[3] Other Asian and Pacific Islander includes Korean, Filipino, Japanese, Laotian, Thai, Pakistani and Hawaiian

[4] West Indian and Caribbean include Jamaican and Barbadian

[5] African includes Nigerian and other African

[6] Middle Easterner includes Lebanese, Iranian and Israeli

Chapter 5

Prevalence of Birth Defects by Region

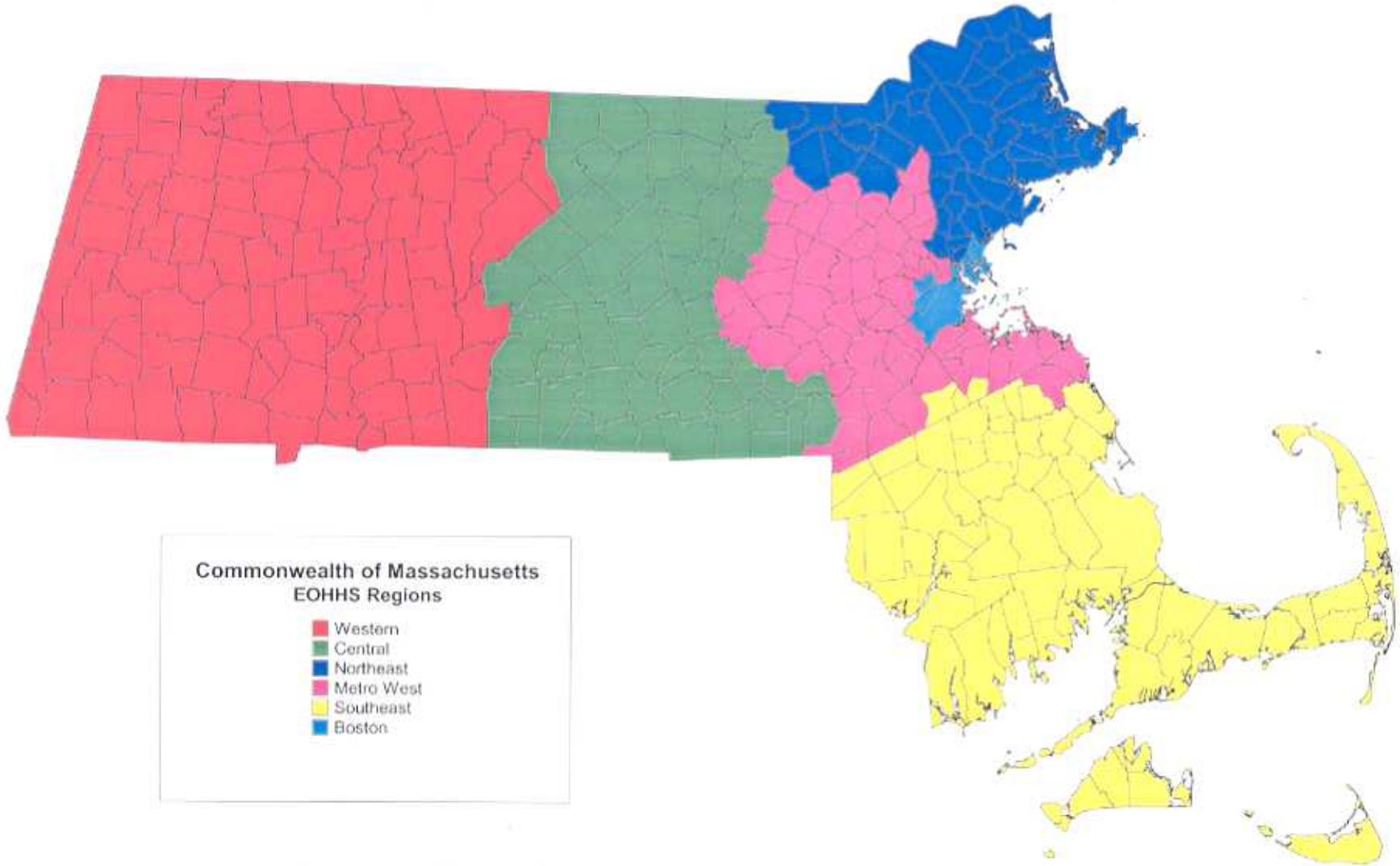
Prevalence of Birth Defects by Region

The map in Figure 7 presents the six regions of the state based on the Massachusetts Executive Office of Health and Human Services (EOHHS) designated areas. Table 13 shows the overall distribution of birth defects across these EOHHS regions. The prevalence rates per 10,000 live births are 147.3 in the West, 113.9 in the Central region, 111.6 in the Northeast, 97.0 in Metro West, 115.3 in Greater Boston and, 86.1 in the Southeast. The lower rate in the Southeast may be due to cases that occurred at Rhode Island hospitals where Massachusetts surveillance was not conducted in 1999. Differences in rates may also be due to differences in the characteristics of the birth populations and in the way birth defects are diagnosed across regions.

There is variation in the most common birth defects by region (see table 14). Common defects (excluding patent ductus arteriosus) in all the regions include septal defects, Down syndrome, cleft lip and cleft palate. Craniosynostosis, obstructive genitourinary defects, transposition of great arteries, and pulmonary valve stenosis and atresia are more common in some regions.

Table 15 shows the prevalence of selected birth defects by region.

Figure 7 Map of EOHHS Regions in Massachusetts



EOHHS = Executive Office of Health and Human Services

Table 13 Overall Distribution of Infants with Birth Defects by Region[1], Massachusetts: 1999

Region	Cases	Rate[2]	95% Confidence Interval
Western	134	147.3	123.4 - 173.3
Central	122	113.9	94.6 - 135.0
Northeast	190	111.6	96.3 - 128.0
Metro West	183	97.0	83.5 - 111.6
Southeast	130	86.1	71.9 - 101.5
Boston	116	115.3	95.2 - 137.2
Total	875	108.2	101.2 - 115.5

[1] Region as defined by the Massachusetts Executive Office of Health and Human Services (EOHHS)

[2] Rate per 10,000 live births

Table 14 Most Common Defects by Region[1,2] Among Live Births, Massachusetts: 1999

Region	Defect	Count	Rate[3]	95 % Confidence Intervals
Western	Cleft Lip with and without Cleft Palate	15	16.5	9.2 - 25.9
	Obstructive Genitourinary Defect	10	11.0	5.2 - 18.9
	Ventricular Septal Defect	9	9.9	4.5 - 17.4
	Hypospadias	8	8.8	3.8 - 15.9
	Trisomy 21 (Down Syndrome)	8	8.8	3.8 - 15.9
	Cleft Palate without Cleft Lip	7	7.7	3.0 - 14.4
Central	Atrial Septal Defect	16	14.9	8.5 - 23.2
	Ventricular Septal Defect	12	11.2	5.8 - 18.4
	Cleft Lip with and without Cleft Palate	8	7.5	3.2 - 13.5
	Trisomy 21 (Down Syndrome)	7	6.5	2.6 - 12.3
	Transposition of Great Arteries	6	5.6	2.0 - 11.0
	Obstructive Genitourinary Defect	6	5.6	2.0 - 11.0
Northeast	Ventricular Septal Defect	20	11.7	7.2 - 17.5
	Cleft Lip with and without Cleft Palate	19	11.2	6.7 - 16.7
	Atrial Septal Defect	17	10.0	5.8 - 15.3
	Trisomy 21 (Down Syndrome)	13	7.6	4.0 - 12.3
	Hypospadias	11	6.5	3.2 - 10.8
Metro West	Atrial Septal Defect	22	11.7	7.3 - 17.0
	Trisomy 21 (Down Syndrome)	15	8.0	4.4 - 12.5
	Pulmonary Valve Atresia and Stenosis	14	7.4	4.0 - 11.8
	Cleft Lip with and without Cleft Palate	14	7.4	4.0 - 11.8
	Hypospadias	12	6.4	3.3 - 10.5
Southeast	Atrial Septal Defect	19	12.6	7.6 - 18.9
	Trisomy 21 (Down Syndrome)	12	7.9	4.1 - 13.1
	Cleft Palate without Cleft Lip	10	6.6	3.2 - 11.4
	Craniosynostosis	8	5.3	2.3 - 9.6
	Obstructive Genitourinary Defect	8	5.3	2.3 - 9.6

Table 14 Most Common Defects by Region[1,2] Among Live Births, Massachusetts: 1999

Region	Defect	Count	Rate[3]	95 % Confidence Intervals
Boston	Ventricular Septal Defect	8	5.3	2.3 - 9.6
	Atrial Septal Defect	11	10.9	5.4 - 14.8
	Hypospadias	10	9.9	4.7 - 13.6
	Trisomy 21 (Down Syndrome)	10	9.9	4.7 - 13.6
	Ventricular Septal Defect	10	9.9	4.7 - 13.6
	Coarctation of Aorta	8	8.0	3.4 - 11.3

[1] Excludes Patent Ductus Arteriosus (PDA) due to the high number of cases and the mild severity of the majority of these cases.

[2] Region as defined by the Massachusetts Executive Office of Health and Human Services (EOHHS)

[3] Rate per 10,000 live births

Table 15 (cont.) Prevalence of Birth Defect Systems by Region for[1] Live Births, Massachusetts: 1999

System[2]	Region	Cases	Rate[3]	95% Confidence Interval
CENTRAL NERVOUS SYSTEM	Western	13	14.3	7.6 - 23.1
	Central	17	15.9	9.2 - 24.3
	Northeast	19	11.2	6.7 - 16.7
	Metro West	18	9.5	5.6 - 14.5
	Southeast	13	8.6	4.6 - 13.9
	Boston	9	8.9	4.1 - 15.7
Total Central Nervous System Cases		89		
EYE	Western	1	1.1	0.0 - 4.3
	Central	5	4.7	1.5 - 9.7
	Northeast	5	2.9	0.9 - 6.1
	Metro West	8	4.2	1.8 - 7.7
	Southeast	1	0.7	0.0 - 2.6
	Boston	4	4.0	1.0 - 8.8
Total Eye Cases		24		
EAR	Western	3	3.3	0.6 - 8.1
	Central	2	1.9	0.2 - 5.4
	Northeast	3	1.8	0.3 - 4.3
	Metro West	3	1.6	0.3 - 3.9
	Southeast	1	0.7	0.0 - 2.6
	Boston	1	1.0	0.0 - 3.9
Total Ear Cases		13		
CARDIOVASCULAR/RESPIRATORY	Western	36	39.6	27.7 - 53.6
	Central	47	43.9	32.2 - 57.3
	Northeast	61	35.8	27.4 - 45.4
	Metro West	61	32.3	24.7 - 41.0
	Southeast	47	31.1	22.9 - 40.6
	Boston	48	47.7	35.2 - 62.1
Total Cardiovascular/Respiratory Cases		300		
OROFACIAL				
	Western	25	27.5	17.8 - 39.3
	Central	12	11.2	5.8 - 18.4
	Northeast	31	18.2	12.4 - 25.2
	Metro West	26	13.8	9.0 - 19.6
	Southeast	16	10.6	6.0 - 16.4
	Boston	14	13.9	7.6 - 22.2
Total Orofacial Cases		124		
GASTROINTESTINAL	Western	16	17.6	10.0 - 27.3
	Central	17	15.9	9.2 - 24.3
	Northeast	21	12.3	7.6 - 18.2
	Metro West	13	6.9	3.7 - 11.1
	Southeast	10	6.6	3.2 - 11.4
	Boston	10	9.9	4.7 - 17.0
Total Gastrointestinal System Cases		87		

Table 15 (cont.) Prevalence of Birth Defect Systems by Region for[1] Live Births, Massachusetts: 1999

System[2]	Region	Cases	Rate[3]	95% Confidence Interval
GENITOURINARY	Western	23	25.3	16.0 - 36.7
	Central	11	10.3	5.1 - 17.2
	Northeast	27	15.9	10.4 - 22.4
	Metro West	24	12.7	8.1 - 18.3
	Southeast	16	10.6	4.8 - 13.2
	Boston	15	14.9	8.3 - 23.4
Total Genitourinary System Cases		116		
MUSCULOSKELETAL	Western	34	37.4	25.9 - 51.0
	Central	28	26.1	17.4 - 36.7
	Northeast	44	25.8	18.8 - 34.0
	Metro West	49	26.0	19.2 - 33.8
	Southeast	41	27.1	19.5 - 36.1
	Boston	31	30.8	20.9 - 42.6
Total Musculoskeletal System Cases		227		
CHROMOSOMAL/SYNDROMES	Western	20	22.0	13.4 - 32.7
	Central	23	21.5	13.6 - 31.1
	Northeast	28	16.4	10.9 - 23.1
	Metro West	34	18.0	12.5 - 24.6
	Southeast	23	15.2	9.6 - 22.1
	Boston	17	16.9	9.8 - 25.9
Total Chromosomal/Syndromes Cases		145		
OTHER	Western	1	1.1	0.0 - 4.3
	Central	0	0.0	
	Northeast	3	1.8	0.3 - 4.3
	Metro West	3	1.6	0.3 - 3.9
	Southeast	1	0.7	0.0 - 2.6
	Boston	3	3.0	0.6 - 7.3
Total Other Cases		11		
Total Cases		875		
Total Defects		1136		

[1] Region as defined by the Massachusetts Executive Office of Health and Human Services (EOHHS)

[2] Cases can be included in more than one system category. However, cases are counted once within a system category.

[3] Rate per 10,000 live births

Chapter 6

Prevalence of Birth Defects by Severity

Prevalence of Birth Defects by Severity

Cases with birth defects were categorized by their level of severity (see Table 16). A severity scale was developed by the Center in collaboration with our partners at Boston University and the Massachusetts General Hospital. This scale was based on the usual outcome for a specific birth defect including its typical compatibility with survival, the need for immediate treatment, the need for long-term care, and the amenability of the defect to correction. A severity score was assigned to each case based on the most severe defect for that infant/fetus. Specific severity category definitions used in this report are as follows:

SEVERITY CATEGORIES	PERCENTAGE OF BIRTH DEFECTS CASES
Always Severe, Usually Incompatible with Life	3%
Severe Need Immediate Treatment Probable Long Term Needs	23%
Moderately Severe Usually Correctable	64%
Mild Minimal or No Correction Needed	10%

Figure 8 shows the distribution of birth defects cases by severity groups

Three percent of cases with birth defects classified as “always severe” did not survive the neonatal period. This percentage is an underestimate of these most severe cases due to limitations of the data, and because it is estimated that up to 80% of anencephaly cases and 50% of any neural tube defect may be electively terminated after prenatal diagnosis (Cragan 2000). These defects led to death in over 80% of cases in the immediate neonatal period for Massachusetts.

Twenty-three percent of cases are affected with a severe birth defect. These cases typically require intensive medical care and planning for continuing care, and experience long-term disability.

Moderately severe birth defects comprise 64% of the total cases. All of these children need medical follow up, and many need surgeries and extensive treatment.

Mild birth defects comprise 10% of the cases. Within the classification of “mild severity”, there is some degree of variation. For example, children with microtia (small ears) may need corrective surgery, hearing evaluations and family support services.

Table 16 Distribution of Birth Defects by Severity, Massachusetts: 1999

Always Severe	
Usually Incompatible with Life	
Anencephaly	7
Bilateral Renal Agenesis	3
Thanatophoric Dysplasia	1
Trisomy 13	2
Trisomy 18	11
TOTAL	24

Severe, Need Immediate Treatment	
Probably Long Term Needs	
Achondroplasia	2
Ambiguous Genitalia	5
Amniotic Band Syndrome	4
Aniridia	1
Anophthalmia	0
Anotia	1
Beckwith-Wiedemann	4
Bladder Exstrophy	1
Biliary Atresia	3
Cloacal Exstrophy	0
Coloboma	9
Common Truncus	1
DiGeorge	6
Encephalocele	4
Endocardial Cushion Defect	32
Epidermolysis Bullosa	2
Holoprosencephaly	6
Hypoplastic Left Heart Syndrome	12
Limb Reductions	22
Microcephaly	19
Osteogenesis Imperfecta	2
Prader Willi Syndrome	3
Sacral Agenesis	0
Spina Bifida	11
Total Anomalous Pulmonary Venous Return	5
Transposition of Great Arteries	21
Trisomy 21	65
TOTAL	241

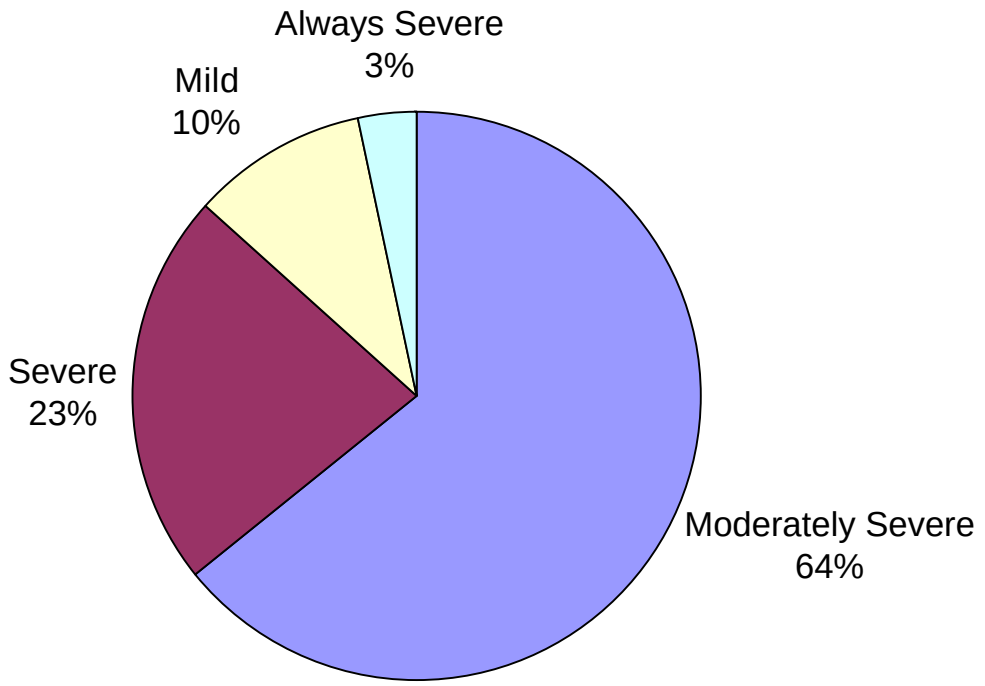
Moderately Severe	
Usually Correctable	
Aortic Valve Stenosis	12
Atrial Septal Defect	91
Choanal Atresia	2
Cleft Lip and Cleft Palate	116
Coarctation of Aorta	25
Congenital Cataract	11
Craniosynostosis	14
Diaphragmatic Hernia	16
Esophageal Atresia/Tracheosophageal Fistula	20
Ebstein Anomaly	4
Gastroschisis	14
Heterotaxy	12
Hirschsprung Disease	10
Hydrocephaly	22
Hypospadias	48
Intestinal and Rectal Atresia / Stenosis	39
Klinefelter Syndrome	9
Obstructive Genitourinary	67
Omphalocele	11
Partial Anomalous Pulmonary Venous Return	4
Pulmonary Valve Atresia / Stenosis	33
Single Ventricle	3
Tethered Cord	5
Tetralogy of Fallot	24
Tricuspid Valve Atresia / Stenosis	7
Turner Syndrome	7
Ventricular Septal Defect	68
TOTAL	694

Mild, Minimal or No Correction Needed	
Microphthalmia	4
Microtia	10
Patent Ductus Arteriosus	103
Poly/Syndactyly	96
X-linked ichthyosis	2
TOTAL	215

N = 1,174

Defects in "other" groups within defect categories were exlcuded from this analysis.

Figure 8 Distribution of Cases with Birth Defects by Severity Groups, Massachusetts: 1999



N=723
Cases with birth defects in "other" groups within defect categories were excluded from this analysis.

Appendices

Technical Notes

Definitions

1999 Denominators Used in Calculating Rates

Birth Defects Codes and Exclusions by Defect Category

All ICD9/BPA Codes with Counts-Live Births and Stillbirths

Glossary

References

Technical Notes

Data Sources

Surveillance records were matched to birth certificate records to gain supplemental information or to verify information on the cases. All but one record was matched. That case was an adopted infant and was excluded from the data analysis. Birth certificate data were used as the source of information for mother's date of birth, race/ethnicity, and town of residence for the regional analysis. Surveillance records provided all diagnostic and the remaining demographic information.

Prevalence, Rates and Confidence Intervals

Prevalence is defined as the number of individuals with a disease or condition at a specific time divided by the number of individuals at risk. The numerator is the number of cases of birth defects. Since the preferred denominator is all pregnancies and since the number of pregnancies cannot be determined, the number of total births is normally used as an approximation.

The rates provided in the tables are estimations of the proportion of infants born with birth defects. This rate is usually expressed as birth defect births per 10,000 births and is calculated by the formula:

Cases/total number live births x 10,000

Since fetal deaths are included in the numerator but not in the denominator, the result is technically a ratio, not a rate. This is consistent with how other birth defects surveillance systems calculate their rates. Because the number of fetal deaths is so small, the inclusion of fetal deaths in the denominator does not substantially change the ratio.

The confidence interval (CI) is a method of assessing the magnitude and stability of a rate or ratio. The CI represents a range of values that has a 95% probability of including the true rate or ratio. Observed rates are subject to statistical variation. Thus, even if the underlying risk of an infant being born with a birth defect is identical in two subpopulations, the observed rates for the subpopulations may differ because of random variation. The confidence interval describes the precision of the observed rate as an estimate of the underlying risk of being born with a birth defect, with a wider interval indicating less certainty about this estimate. The width of the interval reflects the size of the subpopulation and the number of cases of birth defects. Smaller subpopulations with fewer defects lead to wider confidence intervals. The 95% confidence intervals used in the report are based on the Poisson distribution.

Assignment of Race/Ethnicity

This method follows the recommendation of the National Center for Health Statistics of classifying births according to the self-reported race/ethnicity of the mother. The Massachusetts birth certificate records mother's race and ethnicity, including Hispanic ethnicity and was used to more accurately calculate Hispanic specific rates of birth

defect prevalence. Race / ethnicity is a self-reported item and is subject to the usual limitations of this type of information.

Calculation of 1999 Dollars
1999 dollars were calculated from the Gross Domestic Product Deflator Inflation Index. An inflation calculator for adjusting costs from one year to another using the Gross Domestic Product (GDP) Deflator Inflation Index of 1.1391 representing the inflation from 1992 to 1999. This inflation calculator is based on the inflation rate during the US Government Fiscal Year, which begins on October 1 and ends on September 30. http://www.jsc.nasa.gov/bu2/inflateGDP.html

Definitions
These definitions are derived from the Massachusetts Births, 1999 Report.

Birthweight
The weight of an infant recorded at the time of delivery. It may be recorded in either pounds/ounces or grams.

1 pound = 453.6 grams
1,000 grams = 2 pounds and 3 ounces

Infant
A child whose age is less than one year (365 days).

Infant Death
Death of a child whose age is less than one year.

Live Birth
A live birth is any infant who breathes or shows any other evidence of life (such as beating of the heart, pulsation of the umbilical cord, or definite movement of voluntary muscles) after separation from the mother's uterus, regardless of the duration of gestation.

Neonatal
Infant under 28 days of age.

Neonatal Death
Death of a child whose age is less than 28 days.

Plurality
The number of births to a woman from the same pregnancy. A singleton is the birth of one infant; multiple birth represents more than one infant.

Resident Birth

The birth of an infant whose mother reports that her usual place of residence is in Massachusetts. In Massachusetts, a resident is a person with a permanent address in one of the 351 cities or towns.

Stillbirth

A stillbirth is the birth of a fetus at greater than or equal to 20 weeks gestation, or with a weight of at least 350 grams.

1999 Populations Used in Calculating Rates

		Numbers of Live Births
Overall		80,866
By Mother's Age	<20	5,588
	20-24	11,813
	25-29	20,382
	30-34	26,330
	35+	16,753
By Infant's sex	Male	41,435
	Female	39,430
By Plurality	Singleton	77,473
	Multiple Birth	3,393
By Region	West	9,098
	Central	10,710
	Northeast	17,030
	Metrowest	18,859
	Southeast	15,104
	Greater Boston	10,065
By Mother's Race/Ethnicity	White	60,402
	Black	5,844
	Hispanic	8,815
	Asian	4,138
By Mother's Ethnicity	Puerto Rican	4,418
	Dominican	1,555
	Salvadoran	658
	Other Central American	719
	Other Hispanic	1,465
	Chinese	1,179
	Vietnamese	683
	Cambodian	557
	Asian Indian	717
	Other Asian/PI	1,092
	Cape Verdean	783
	Brazilian	799
	Other Portuguese	1,353
	Haitian	1,017
	West Indian/Caribbean	663
	African American	2,885
	African	820
	Middle Eastern	790
	Native American	262
	European	14,543

Birth Defect Codes and Exclusions[1] Massachusetts 1999

Defect	ICD-9 / BPA[2]	NOTES
Central Nervous System		
Anencephaly	740.020-740.100	
Spina Bifida	741.001-741.999 w/o 740.020-740.100	
Hydrocephaly	742.300-742.390 w/o 741.001-741.999	Excludes hydrocephalus due to intraventricular hemorrhage
Encephalocele	742.000-742.090	
Microcephaly	742.100	Include if 2 standard deviations below the mean, adjusted for gestational age and sex
Holoprosencephaly	742.260-742.267	
Tethered Cord	742.580	
Other Central Nervous System	742.210, 230, 240, 250, 280, 400, 410, 420, 900	Excludes Marcus Gunn Syndrome
Eye		
Anophthalmia/Microphthalmia	743.000-743.104	
Congenital Cataract	743.320, 325, 326	
Aniridia	743.420	
Coloboma	743.340, 430, 480, 490, 520, 535, 636	
Other Eye	743.200, 440, 510, 580	Excludes tear duct cysts; blocked tear ducts. Excludes eyelid, lacrimal systems, orbit abnormalities; other specified and unspecified anomalies of the eye
Ear		
Anotia/Microtia	744.010-744.014, 744.210-744.214	Excludes type 1 microtia/macrochelia. Excludes ear tags
Other Ear	744.000	Excludes preauricular sinuses

Birth Defect Codes and Exclusions[1] Massachusetts 1999

Defect	ICD-9 / BPA[2]	NOTES
Cardiovascular/Respiratory		
Common Truncus/Aortic Septal Defects	745.000-745.010	
Transposition of Great Arteries	745.100-745.110, 745.185-745.189 (excluding 745.120)	
Tetralogy of Fallot	745.200, 747.215-217, 747.310	
Ventricular Septal Defect	745.485, 745.487, 745.490 (excluding 745.486 and 745.498)	
Atrial Septal Defect	745.510, 745.599	
Endocardial Cushion Defect	745.600-745.690	
Pulmonary Valve Atresia and Stenosis	746.000-746.010	
Tricuspid Valve Atresia and Stenosis	746.100 (excluding 746.105)	
Ebstein Anomaly	746.200	
Aortic Valve Stenosis	746.300	
Hypoplastic Left Heart Syndrome	746.700	
Patent Ductus Arteriosus	747.000	Excludes isolated PDA with gestational age < 37 weeks
Coarctation of Aorta	747.100-747.190	
Total Anomalous Pulmonary Venous Return	747.420	
Partial Anomalous Pulmonary Venous Return	747.430	
Single Ventricle	745.300-745.380	
Situs Inversus/Heterotaxia	759.300-759.360	

Birth Defect Codes and Exclusions[1] Massachusetts 1999

Defect	ICD-9 / BPA[2]	NOTES
Other Cardiovascular/Respiratory	**745**.120, 500, 590, **746**.080, 090, 400, 480, 500, 505, 600, 800, 830, 480, 490, 881, 882, 885, 900, 995, **747**.200, 210, 220, 230, 250, 270, 280, 320, 380, 640, 480, 810, **748**.400, 410, 500, 510, 520, 580, **759**.330, 340, 390	Excludes unspecified anomalies of circulatory systems and respiratory system; web of larynx; laryngotracheomalacia/tracheomalacia. Excludes single umbilical artery. Excludes peripheral pulmonic stenosis
Orofacial		
Cleft Palate without Cleft Lip	749.001-749.090	Excludes bifid uvula
Cleft Lip with and without Cleft Palate	749.101-749.195, 749.201-749.290	Excludes cleft gum and submucous cleft palate
Choanal Atresia	748.010-748.014	
Pierre Robin Sequence	524.080	
Branchial Cleft/Sinus	744.400, 480	
Other Orofacial	748.000, 748.120, 744.900	
Gastrointestinal		
Esophageal Atresia/Tracheosophageal Fistula	750.300-750.330	
Rectal and Large Intestinal Atresia/Stenosis	751.200-751.240	
Hirschsprung Disease	751.300-751.340	
Biliary Atresia	751.650	
Small Intestinal Atresia	751.100-751.195	
Other Gastrointestinal	**750**.600, 751.520-580, **751**.010, 400, 490, 495, 520, 530, 540, 560, 580, 660, 720, 800	Excludes anal fissure, if isolated. Excludes unspecified anomalies of the digestive system. Excludes tongue tie; protruding tongue, congenital hypertrophic pyloric stenosis and unspecified upper alimentary tract abnormalities
Genitourinary		
Renal Agenesis/Hypoplasia	753.000-753.008	Excludes unilateral renal agenesis/hypoplasia

Birth Defect Codes and Exclusions[1] Massachusetts 1999

Defect	ICD-9 / BPA[2]	NOTES
Bladder Exstrophy	753.500	
Obstructive Genitourinary Defect	753.200-753.290, 753.600-753.690	Excludes isolated diagnosis without surgical intervention and secondary diagnosis without postnatal confirmation
Hypospadias	752.606-752.607, 752.626-752.627	Excludes first degree hypospadias and epispadias
Ambiguous Genitalia	752.790	
Other Genitourinary	**751**.550**, 752**.000, 085, 320, 380, 410, 420, 820, 860, 865, **753**.110, 160, 320, 330, 340, 380, 410, 420, 480, 700, 810, 880	Excludes undescended testicle(s); unspecified anomalies of the genital organs/urinary system.
Musculoskeletal		
Reduction Deformity, Upper Limbs	755.200-755.290	
Reduction Deformity, Lower limbs	755.300-755.390	
Gastroschisis	756.710	
Omphalocele	756.700	
Diaphragmatic Hernia	756.601-756.605, 756.610-756.619	
Achondroplasia	756.430	
Thanatophoric Dysplasia	756.447	
Osteogenesis Imperfecta	756.500	
Craniosynostosis	756.000-756.024	
Other Musculoskeletal	**754**.030, 080, 200, 400, 410, 490, 754.500-590, 754.600, 690, 730, 735, 880, **755**.005, 010, 020, 030, 100, 110, 120, 130, 193, 194, 196, 199, 440, 500, 540, 580, 585, 640, 650, 680, 685, 800, **756**.005, 057, 060, 065, 080, 145, 150, 155, 160, 165, 170, 180, 185, 190, 300, 320, 330, 340, 410, 620, 720, 800, 810, 880	Exclude dislocated hips; other deformities of the feet; specfied/ unspecified musculoskeletal and limb deformities; polydactly postaxial type B; syndactyly of toes 2 and 3; overlapping sutures; supernumerary rib in cervical region. Excludes deviated nasal septum; short neck

Birth Defect Codes and Exclusions[1] Massachusetts 1999

Defect	ICD-9 / BPA[2]	NOTES
Chromosomal/Syndromes		
Trisomy 13 (Patau Syndrome)	758.100-758.190	
Trisomy 21 (Down Syndrome)	758.000-758.090	
Trisomy 18 (Edwards Syndrome)	758.200-758.290	
Turner Syndrome	758.600-758.690	
Klinefelter Syndrome	758.700-758.790	
Prader-Willi/Beckwith-Weidemann	759.870	
DiGeorge Syndrome	279.110	
Other Chromosomal/Syndromes	**758**.300-758.590, 758.800-758.990, **759**.800-759.890, 759.610, 759.500	Excludes balanced translocation in normal individual
Other		
Amniotic Bands	658.800	
Endocrine Anomalies	255.200, 759.240	
Epidermolysis Bullosa	757.330	Excludes minor integument anomalies
X-linked Ichthyosis	757.196	

[1] Other ICD 9 codes and diagnoses outside of the 740.0 - 759.9 range which are also excluded are: Syringomyelia, isolated; inguinal hernia, umbilical hernia, testicular torsion, sacral/pilonidal dimple, tibial torsion, hydroceles, webbing of neck and associated abnormalities, heart murmers without confirmation of a structural defect

[2] Coding scheme derives from International Classification of Diseases (ICD) 9th Revision/British Pediatric Association (BPA), 1979

Selected ICD9/BPA[1] Codes with Counts-Live Births and Stillbirths

BPA Label	BPA Code	# of Defects
Central Nervous System		
Anencephaly	740020	8
Meningomyelocele/myelomeningocele, Highest level, lumbar, Arnold Chiari malformation, unspec. open/closed lesion	741203	3
Lipomeningomyelocele, Highest level, lumbar, Hydrocephalus, other or NOS, open lesion	741343	1
Meningomyelocele/myelomeningocele, Highest level, lumbar, Hydrocephalus	741503	2
Meningomyelocele/myelomeningocele, Highest level, sacral, No mentioned hydrocephalus, open lesion	741704	1
Meningocele, Highest level, lumbar, No mentioned hydrocephalus, closed lesion	741813	2
Lipomeningomyelocele, Highest level, lumbar, No mentioned hydrocephalus, closed lesion	741843	1
Unspecified spina bifida, Highest level unspecified, No mentioned hydrocephalus, unspecified open/closed lesion	741999	1
Hydrocephaly, Anomalies of Aqueduct of Sylvius	742300	4
Dandy-Walker Malformation	742310	9
Hydrocephaly, NOS	742390	10
Encephalocele, Occipital	742000	1
Encephalocele, Other Specified	742080	2
Encephalocele, Parietal	742086	1
Microcephalus	742100	19
Holoprosencephaly, NOS	742260	2
Holoprosencephaly, Alobar	742265	1
Holoprosencephaly, Semilobar	742266	3
Other specified anomalies of spinal cord and membranes	742580	5
Other Central Nervous System		
Anomalies of corpus callosum	742210	19
Anomalies of cerebellum	742230	2
Agyria and lissencephaly	742240	3
Microgyria / polymicrogyria	742250	2
Other specified reduction defect of brain	742280	8
Enlarged brain and or head	742400	2
Porencephaly / porencephalic cysts	742410	1
Cerebral cysts	742420	11
Brain, unspecified anomalies	742900	1
Eye		
Anophthalmos, Left	743001	1
Microphthalmos, Left	743101	1
Microphthalmos, Bilateral	743104	2
Cataract, NOS	743320	10
Cataract, anterior polar	743325	1
Absence of iris / aniridia	743420	1
Coloboma of iris	743430	2
Other specified colobomas and anomalies of anterior segments	743480	2

Selected ICD9/BPA[1] Codes with Counts-Live Births and Stillbirths

BPA Label	BPA Code	# of Defects
Unspecified colobomas and anomalies of anterior eye segments	743490	2
Specified anomalies of optic disc / hypoplastic optic nerve / coloboma of the optic disc	743520	3
Other Eye		
Buphthalmos / congenital glaucoma / hydrophthalmos	743200	1
Other anomalies of iris	743440	2
Specified anomalies of retina / congenital retinal aneurysm	743510	2
Other specified anomalies of posterior segment of eye	743580	1
Ear		
Anotia, Left	744011	1
Microtia, Left	744211	3
Microtia, Right	744212	7
Microtia, Bilateral	744214	2
Other Ear		
Absence or stricture of auditory canal	744000	5
Cardiovascular/Respiratory		
Aortic septal defect / aortopulmonary window	745010	1
Dextro-transposition of great arteries	745100	6
Dextro-transposition of great arteries	745110	7
Double-outlet right ventricle (DORV) with normally related great arteries	745185	1
Double-outlet right ventricle (DORV) with transposed great arteries	745186	4
Double-outlet right ventricle (DORV), NOS	745189	3
Tetralogy of Fallot	745200	22
Pulmonary atresia with VSD (tetralogy of Fallot with pulmonary atresia)	747310	2
Ventricular septal defect, Perimembranous	745485	49
Ventricular septal defect, Malalignment-type	745487	6
Ventricular septal defect, NOS	745490	16
Atrial septal defect, Secundum type (ASD2)	745510	62
Atrial septal defect, NOS	745599	33
Atrial septal defect, primum type (ASD1)	745600	8
Common Atrium	745610	1
Single common atrium	745610	1
Complete atrioventricular canal (CAVC) with ventricular septal defect	745620	7
Complete atrioventricular canal (CAVC)	745630	10
Endocardial cushion defect, Other specified	745680	2
Ventricular septal defect, inflow type	745685	5
Pulmonary valve atresia/intact ventricular septum	746000	6
Pulmonic stenosis, valvar	746010	28
Tricuspid atresia	746100	7
Ebstein Malformation or Anomaly	746200	7

Selected ICD9/BPA[1] Codes with Counts-Live Births and Stillbirths

BPA Label	BPA Code	# of Defects
Aortic stenosis, valvar	746300	12
Hypoplastic left heart syndrome	746700	14
Patent ductus arteriosus (PDA)	747000	103
Coarctation of the aorta, postductal (distal)	747110	1
Coarctation of the aorta, juxtaductal	747120	5
Coarctation of the aorta, NOS	747190	19
Total anomalous pulmonary venous return/connection/drainage	747420	5
Partial anomalous pulmonary venous return/connection/drainage	747430	4
Single ventricle, NOS	745300	1
Single ventricle, Double Inlet Right Ventricle	745320	1
Single ventricle, Other Specified	745380	1
Complete situs inversus w/ dextrocardia	759300	9
Situs inversus w/ levocardia	759310	2
Situs ambiguus, right; right isomerism	759350	2
Other Cardiovascular/Respiratory		
Corrected transposition of great vessels / L-transposition / ventri inversion	745120	2
Patent or persistent foramen ovale (PFO) / Nonclusure of foramen ovale, NOS	745500	55
PFO vs ASD	745590	35
Unspecified anomaly of pulmonary valve	746090	1
Aortic valve insufficiency or regurgitation / congenital bicuspid aortic valve	746400	17
Other specified anomalies of the aortic valves / aortic valve atresia	746480	5
Unspecified anomalies of the aortic valves	746490	1
Congenital mitral stenosis	746500	7
Absence, atresia, or hypoplasia of mitral valve	746505	10
Mitral valve insufficiency or regurgitation, congenital	746600	10
Dextrocardia without situs inversus / dextrocardia with situs solitus	746800	2
Pulmonary infundibular (subvalvular) stenosis	746830	4
Hypoplastic left ventricle	746881	8
Hypoplastic right heart or right ventricle / Uhl's disease	746882	3
Anomalies of coronary artery or sinus	746885	1
"Pulmonic" or pulmonary atresia, stenosis, or hypoplasia, NOS	746995	8
Atresia of aorta / absence of aorta	747200	5
Hypoplasia of aorta / tubular hypoplasia of aorta	747210	20
Supra-aortic stenosis / supravalvular aortic stenosis	747220	1
Persistent right aortic arch	747230	11
Vascular ring (aorta) / double aortic arch / vascular ring compression of trachea	747250	6
Congenital aneurysm of aorta / congenital dilatation of aorta	747270	1
Other specified anomalies of aorta	747280	1
Pulmonary artery stenosis	747320	4

Selected ICD9/BPA[1] Codes with Counts-Live Births and Stillbirths

BPA Label	BPA Code	# of Defects
Other specified anomaly of pulmonary artery / pulmonary artery hypoplasia	747380	2
Persistent left superior vena cava	747410	3
Other specified anomalies of great veins	747480	8
Other anomalies of peripheral arteries / aberrant subclavian artery	747640	3
Other anomalies of cerebral vessels / vein of Galen	747810	1
Single cyst lung	748400	1
Multiple cysts, lung / polycystic lung	748410	2
Agenesis or aplasia of lung	748500	1
Hypoplasia of lung or pulmonary hypoplasia	748510	3
Sequestration of lung	748520	3
Other specified dysplasia of lung / fusion of lobes of lung	748580	1
Situs inversus abdominis	759330	2
Orofacial		
Cleft hard palate, Bilateral	749010	4
Cleft hard palate, NOS	749030	4
Cleft soft palate, Unilateral, Right	749042	1
Cleft soft palate, Unilateral, Side Unk	749043	1
Cleft soft palate, Bilateral	749050	4
Cleft soft palate, Central	749060	4
Cleft soft palate, NOS	749070	19
Cleft palate, NOS	749090	12
Cleft lip, Unilateral, Left	749101	14
Cleft lip, Unilateral, Right	749102	6
Cleft lip, Bilateral	749110	2
Cleft lip, NOS	749195	2
Cleft lip and palate, Unilateral cleft lip, Left	749201	17
Cleft lip and palate, Unilateral cleft lip, Right	749202	17
Cleft lip and palate, Bilateral cleft lip	749210	8
Cleft lip and palate, NOS	749290	3
Choanal atresia, Unilateral, Side Unk	748013	1
Choanal atresia, Bilateral	748014	1
Pierre Robin sequence	524080	15
Brachial cleft, sinus, fistula, cyst, or pit	744400	6
Other brachial cleft anomalies / dermal sinus of head	744480	2
Other Orofacial		
Congenital anomaly of neck	744900	2
Choanal atresia / atresia of nares	748000	4
Fissured, notched, or cleft nose	748120	1
Gastrointestinal		

Selected ICD9/BPA[1] Codes with Counts-Live Births and Stillbirths

BPA Label	BPA Code	# of Defects
Esophageal atresia without TE fistula	750300	3
Esophageal atresia with TE fistula	750310	17
Tracheoesophageal fistula without mention of esophageal atresia	750320	1
Tracheoesophageal fistula, "H" type	750325	2
Rectal atresia/stenosis with fistula	751210	1
Rectal atresia/stenosis without mention of fistula	751220	2
Anal atresia with fistula	751230	5
Anal atresia without mention of fistula	751240	11
Long-segment Hirschsprung's Disease	751310	2
Hirschsprung's Disease, NOS	751330	8
Biliary atresia, extrahepatic or NOS	751650	3
Intestinal atresia/stenosis, Duodenum	751100	11
Intestinal atresia/stenosis, Jejunum	751110	8
Intestinal atresia/stenosis, Ileum	751120	4
Other Gastrointestinal		
Meckel's diverticulum	751010	2
Malrotation of cecum and/or colon	751400	3
Other specified and unspecified malrotation	751490	5
Malrotation of small intestine alone	751495	2
Microcolon	751520	2
Ectopic (displaced) anus	751530	3
Congenital anal fistula	751540	1
Duodenal web	751560	1
Other specified anomalies of intestine / Rectal fissures	751580	1
Choledochal cysts	751660	1
Annular pancreas	751720	2
Genitourinary		
Renal agenesis, bilateral	753000	4
Bladder exstrophy	753500	1
Congenital hydronephrosis / pyelocaliectasis	753200	34
Atresia, stricture, or stenosis of ureter / ureteropelvic junction obstruction or stenosis /ureterovesical junction obstruction or stenosis / hypoplastic ureter	753210	18
Megaloureter, NOS / hydroureter	753220	9
Congenital posterior urethral valves or posterior urethral obstruction	753600	4
Other atresia, or stenosis of bladder neck	753610	1
Obstruction, atresia or stenosis of anterior urethra	753620	1
Hypospadias, Second Degree	752606	21
Hypospadias, Third Degree	752607	13
Hypospadias, Second Degree with Chordee	752626	9
Hypospadias, Third Degree with Chordee	752627	6

Selected ICD9/BPA[1] Codes with Counts-Live Births and Stillbirths

BPA Label	BPA Code	# of Defects
Indeterminate sex, NOS / ambiguous genitalia	752790	7
Other Genitourinary		
Absence or agenesis of ovaries	752000	1
Multiple ovarian cysts	752085	1
Fistulae connecting uterus with digestive or urinary tract/ uterointestinal fistula / uterovesical fistula	752320	1
Other anomalies of uterus / bicornuate uterus / unicornis uterus	752380	1
Absence or atresia of vagina, complete or partial	752410	1
Congenital rectovaginal fistula	752420	1
Other anomalies of testis and scrotum / polyorchidism / bifid scrotum	752820	2
Other anomalies of penis / absent or hooded foreskin	752860	2
Small penis / hypoplastic penis / micropenis	752865	7
Polycystic kidneys, infantile type	753110	1
Multicystic renal dysplasia / multicystic kidney	753160	4
Lobulated, fused, or horseshoe kidney	753320	4
Ectopic kidney / pelvic kidney	753330	2
Accessory ureter / double ureter / duplex collecting system	753410	9
Ectopic ureter	753420	3
Other specified anomalies of ureter / ureterocele	753480	5
Patent urachus	753700	1
Other specified anomalies of bladder and urethra	753880	1
Musculoskeletal		
Absence of hand or fingers, Laterality Unk	755245	1
Absence of hand or fingers, Left	755246	3
Absence of hand or fingers, Right	755247	4
Absence of hand or fingers, Bilateral	755249	1
Longitudinal deficiency of arm, NOS, Right	755252	2
Split-Hand, Bilateral	755259	2
Thumb only missing or hypoplastic, Left	755261	1
Thumb only missing or hypoplastic, Right	755262	1
Thumb only missing or hypoplastic, Bilateral	755264	1
Absence of lower leg only (thigh and foot present), Left	755336	1
Absence of foot or toes, Laterality Unk	755345	1
Absence of foot or toes, Left	755346	1
Absence of foot or toes, Right	755347	1
Split-Foot, Left	755356	1
Split-Foot, Bilateral	755359	2
Tibial aplasia/hypoplasia, Left	755366	1
Tibial aplasia/hypoplasia, Bilateral	755369	1
Fibular aplasia/hypoplasia, Right	755372	1

Selected ICD9/BPA[1] Codes with Counts-Live Births and Stillbirths

BPA Label	BPA Code	# of Defects
Gastroschisis	756710	14
Omphalocele	756700	13
Diaphragmatic hernia, NOS	756600	1
Diaphragmatic hernia, NOS	756601	12
Diaphragmatic hernia, NOS	756602	1
Diaphragmatic hernia, NOS	756604	1
Diaphragmatic hernia, Bochdalek, Left	756611	1
Diaphragmatic hernia, Morgagni, Right	756617	1
Achondroplastic dwarfism	756430	2
Thanatophoric dwarfism	756447	1
Osteogenesis imperfecta	756500	2
Craniosynostosis, Unspecified Type, Laterality Unk	756000	1
Craniosynostosis, Sagittal	756005	6
Craniosynostosis, Metopic	756006	2
Craniosynostosis, Coronal, Left	756011	1
Craniosynostosis, Coronal, Right	756012	2
Craniosynostosis, Coronal, Bilateral	756014	1
Craniosynostosis, Lambdoidal, Right	756022	1
Other Musculoskeletal		
Dolichocephaly	754030	2
Other specified skull deformity	754080	1
Congenital postural scoliosis	754200	5
Bowing, femur	754400	1
Deformity of leg, NOS	754490	1
Talipes equinovarus / clubfoot equinovarus	754500	20
Metatarsus varus or adductus	754520	8
Talipes calcaneovalgus	754600	3
Unspecified valgus deformities of foot	754690	1
Clubfoot, NOS / talipes, NOS	754730	17
Accessory fingers / postaxial polydactyly, Type A	755005	10
Accessory thumbs (preaxial polydactyly)	755010	22
Accessory toes (postaxial)	755020	12
Accessory big toe (preaxial)	755030	5
Fused fingers	755100	4
Webbed fingers	755110	10
Fused toes	755120	7
Webbed toes //Webbing between the second and third toes	755130	2
Unspecified (webbed vs. fused) syndactyly, thumb and / or fingers, NOS	755193	8
Unspecified syndactyly toes	755194	14

Selected ICD9/BPA[1] Codes with Counts-Live Births and Stillbirths

BPA Label	BPA Code	# of Defects
Unspecified syndactyly toes, laterality not spec	755196	1
Unspecified (webbed vs. fused) syndactyly, digits not known	755199	1
Absent digits, NOS	755440	1
Anomalies of fingers	755500	14
Anomalies of elbow and upper arm	755540	2
Other specified anomalies of upper limb	755580	2
Hypoplasia of upper limb / hypoplasia of fingers, hands, or arms	755585	7
Anomalies of knee / hyperextended knee	755640	1
Anomalies of upper leg / anteversion of femur	755650	1
Other specified anomalies of lower limb / hyperextended legs / shortening of legs	755680	2
Hypoplasia of lower limb / hypoplasia of toes, feet, legs	755685	2
Arthrogryposis multiplex congenita / distal arthrogryposis syndrome	755800	3
Acrocephalosyndactyly types I or II / Apert syndrome	756055	1
Other specified acrocephalosyndactylies	756057	1
Goldenhar syndrome / oculoauriculovertebral dysplasia	756060	2
Hemifacial microsomia	756065	2
Other specified skull and face bone anomalies	756080	3
Hemivertebrae (cervical)	756145	1
Hemivertebrae of thoracic vertebrae	756155	7
Anomalies of lumbar vertebrae	756160	1
Hemivertebrae of lumbar vertebrae	756165	2
Sacrococcygeal anomalies / agenesis of sacrum	756170	1
Other specified vertebral anomalies	756180	3
Absence of ribs	756300	2
Fused ribs	756320	3
Extra ribs	756330	3
Other anomalies of ribs	756340	1
Chondrodysplasia / Ollier syndrome / enchondromatosis	756410	2
Eventration of diaphragm	756620	1
Prune belly syndrome	756720	1
Poland syndrome or anomaly	756800	4
Chromosomal/Syndromes		
Patau syndrome with karyotype trisomy 13	758100	2
Down syndrome with karyotype trisomy 21	758000	63
Translocation trisomy - duplication of a number_21 chromosome	758020	3
Mosaic Down syndrome	758040	1
Edwards syndrome with karyotype trisomy 18	758200	16
Turner phenotype with karyotype 45, X [XO]	758600	4
Turner phenotype with variant karyotypes	758610	5

Selected ICD9/BPA[1] Codes with Counts-Live Births and Stillbirths

BPA Label	BPA Code	# of Defects
Klinefelter's phenotype with karyotype 47, XXY	758700	6
Klinefelter syndrome, NOS	758790	3
DiGeorge syndrome	279110	6
Other Chromosomal/Syndromes		
Wolff-Hirschorn syndrome with karyotype deletion of_4 or a B-group chromosome, NOS	758320	1
Deletion of short arm of E/deletion short arm of 17 or 18	758350	1
Other loss of autosomal material / microdeletion	758380	2
Other trisomy C syndromes /trisomy	758510	1
Other total trisomy syndromes	758520	1
Partial trisomy syndromes	758530	3
Other translocations	758540	3
Polyploidy	758585	1
Triploidy	758586	1
XYY, male /_47XYY / mosaic XYY male	758840	2
XXX female /_47XXX	758850	1
Mosaicism, NOS	758900	2
Unspecified anomaly of chromosome(s)	758990	3
Tuberous sclerosis / Bourneville's disease / epiloia	759500	1
Encephalocutaneous angiomatosis / Kalischer's disease / Sturge-Weber syndrome	759610	1
Congenital malformation syndromes affecting facial appearance	759800	2
Congenital malformation syndromes associated with short stature	759820	3
Congenital malformation syndromes involving limbs	759840	4
Congenital malformation syndromes with other skeletal changes	759860	1
Congenital malformation syndromes with metabolic disturbances	759870	7
Other specified anomalies / hemihypertrophy / Meckel-Gruber syndrome	759890	9
Other		
Amniotic band sequence	658800	6
Adrenogenital syndrome / adrenal hyperplasia	255200	2
Anomalies of thymus / absent thymus	759240	2
Epidermolysis bullosa	757330	2
X-linked ichthyosis	757196	2

[1] Coding scheme derives from International Classification of Diseases (ICD) 9th Revision /British Pediatric Association (BPA), 1979

Glossary of Birth Defects Terms[1]

Agenesis: Absence of part(s) of the body.

Agenesis, aplasia, or hypoplasia of the lung: The absence or incomplete development of a lung or lung tissue.

Anencephaly: Congenital absence of the skull, with cerebral hemispheres completely missing or reduced to small masses attached to the base of the skull. Anencephaly is not compatible with life.

Aniridia: The complete absence of the iris of the eye or a defect of the iris.

Anophthalmia: A developmental defect characterized by complete absence of the eyes, or by the presence of vestigial eyes.

Anotia: A congenital absence of one or both ears.

Aortic valve stenosis: A cardiac anomaly characterized by a narrowing or stricture of the aortic valve.

Atresia / Imperforation: Absence or closure of a normal opening.

Atrial Septal Defect: A congenital cardiac malformation in which there are one or several openings in the atrial septum (wall between the right and left atria). Most common type is called ostium secundum defect, although there are other locations.

Biliary atresia: A congenital absence or underdevelopment of one or more of the ducts in the biliary tract.

Bladder exstrophy: Incomplete closure of the anterior wall of the bladder and the abdominal cavity. The upper urinary tract is generally normal. Often associated with anorectal and genital malformations.

Branchial cleft: A defect found in the neck or just below the collarbone (clavicle) that may be visible as pits in the skin, projections of tissue, or lumps. These defects appear early during development of the embryo when tissue of the branchial cleft (the area in the neck and collarbone) fail to develop in a normal manner. The cyst may appear on one or both sides. They do not spontaneously resolve, and often become infected.

Cataract: An opacity (clouding) of the lens of the eye.

Choanal atresia or stenosis: A congenital anomaly in which a bony or membranous formation blocks the passageway between the nose and the pharynx. This defect is usually repaired surgically after birth.

Cleft lip: The congenital failure of the fetal components of the lip to fuse or join, forming a groove or fissure in the lip. Infants with this condition can have difficulty feeding and

may use assistive devices for feeding. This condition is corrected when the infant can tolerate surgery.
Cleft palate: The congenital failure of the palate to fuse properly forming a grooved depression or fissure in the roof of the mouth. This defect varies in degree of severity. The fissure can extend into the hard and soft palate and into the nasal cavities. Infants with this condition have difficulty feeding, and may use assistive devices for feeding. Surgical correction is begun as soon as possible. Children with cleft palates are at high risk for hearing problems due to ear infections.

Coarctation of the aorta: Localized narrowing of the aorta. This condition can vary from mild to severe.

Common truncus arteriosus: A congenital heart defect in which the common arterial trunk fails to divide into pulmonary artery and aorta.

Congenital: Existing at or dating from birth although the defect may not be recognized at the time of birth.

Corpus callosum: The area of the brain which connects the two cerebral hemispheres.

Craniosynostosis: A premature ossification (closing) of the cranial sutures before birth or soon after birth. This condition is occasionally associated with other skeletal defects. If no surgical correction is made, the growth of the skull is inhibited, and the head is deformed.

Diaphragmatic hernia: A failure of the diaphragm to form completely, leaving a hole. Abdominal organs can protrude through the hole into the chest cavity and interfere with development of the heart and lungs. Usually life-threatening and requires emergent surgery.

Down syndrome (Trisomy 21): The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. Down syndrome is characterized by moderate to severe mental retardation. Many infants have congenital heart disease.

Ebstein anomaly: A congenital heart defect in which the tricuspid valve is displaced downward into the right ventricle.

Edwards syndrome: See Trisomy 18**.**

Encephalocele: Herniation of the brain through a defect in the skull.

Endocardial cushion defect: In the complete form, a septal defect involving both the upper chambers (atria, atrial septal defect) and lower chambers (ventricles, ventricular septal defect) such that there is a single large atrioventricular septal defect. There are incomplete forms as well.

Esophageal stenosis or atresia: A narrowing or incomplete formation of the esophagus. Usually a surgical emergency. Frequently associated with a Tracheoesophageal Fistula.

Fistula: An abnormal passage from an internal organ to the body surface or between two internal organs or structures.

Gastroschisis: A congenital opening of the abdominal wall with protrusion of the intestines. This condition is surgically treated.

Hirschsprung disease: The congenital absence of autonomic ganglia (nerves controlling involuntary and reflexive movement) in the muscles of the colon. This results in immobility of the intestines and may cause obstruction or stretching of the intestines. This condition is repaired surgically in early childhood by the removal of the affected portion of the intestine.

Holoprosencephaly: Failure of the brain to develop into two equal halves, There is structural abnormality of the brain. There may be associated midline facial defects.

Hydrocephalus: The abnormal accumulation of fluid within the spaces of the brain.

Hyperplasia: Overgrowth characterized by an increase in the number of cells of tissue.

Hypoplasia: A condition of arrested development in which an organ or part remains below the normal size or in an immature state.

Hypoplastic left heart syndrome: Atresia, or marked hypoplasia, of the aortic valve, atresia or marked hypoplasia for the mitral valve, with hypoplasia of the ascending aorta and underdevelopment of the left ventricle.

Hypospadias: A congenital defect in which the urinary meatus (urinary outlet) is on the underside of the penis or on the perineum (area between the genitals and the anus). The urinary sphincters are not defective so incontinence does not occur. The condition may be surgically corrected if needed for cosmetic, urologic, or reproductive reasons.

Limb defects: See Reduction defects.

Meninges: Membranes that cover the brain and spinal cord.

Microcephaly: Congenital smallness of the head, with corresponding smallness of the brain.

Microphthalmia: The congenital abnormal smallness of one or both eyes. Can occur in the presence of other ocular defects.

Microtia: A small or maldeveloped external ear and atretic or stenotic external auditory canal.

Mosaic: In genetics, this refers to an individual organism that has two or more kinds of genetically different cell types. The degree of abnormality depends on the type of tissue containing affected cells. Individuals may vary from near normal to full manifestation of the genetic syndrome. Can occur in any chromosome abnormality syndrome.

Neural tube defect: A defect resulting from failure of the neural tube to close in the first month of pregnancy. The major conditions include anencephaly, spina bifida, and encephalocele.

Obstructive genitourinary defect: Stenosis or atresia of the urinary tract at any level. Severity of the defect depends largely upon the level of the obstruction. Urine accumulates behind the obstruction.

Omphalocele: The protrusion of intestines into the umbilicus. The defect is usually closed surgically soon after birth.

Patau Syndrome: See Trisomy 13.

Patent ductus arteriosus: A blood vessel between the pulmonary artery and the aorta. This is normal in fetal life, but can cause problems after birth, particularly in premature infants.

Pulmonary artery anomaly: Abnormality in the formation of the pulmonary artery such as stenosis or atresia.

Pulmonary valve atresia or stenosis: A congenital heart condition characterized by absence or constriction of the pulmonary valve. Pulmonary atresia can occur with an intact ventricular septum or with a VSD. In the latter situation, it is more properly called Tetralogy of Fallot with pulmonary atresia.

Reduction defects of the lower limbs: The congenital absence of a portion of the lower limb. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (for example, a missing tibia and great toe).

Reduction defects of the upper limbs: The congenital absence of a portion of the upper limb. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (for example, a missing radius and thumb).

Renal agenesis or dysgenesis: The failure, or deviation, of embryonic development of the kidney.

Spina bifida: A neural tube defect resulting from failure of the spinal neural tube to close. The spinal cord and/or meninges may or may not protrude. This usually results in damage to the spinal cord with paralysis of the involved limbs. Includes myelomeningocele (involving both spinal cord and meninges) and meningocele (involving just the meninges).

Stenosis: A narrowing or constriction of the diameter of a bodily passage or orifice.

Stenosis or atresia of large intestine, rectum and anus: The absence, closure or constriction of the large intestine, rectum or anus. Can be surgically corrected or bypassed.

Stenosis or atresia of the small intestine: A narrowing or incomplete formation of the small intestine obstructing movement through the digestive tract.

Tetralogy of Fallot: A congenital cardiac anomaly consisting of four defects: ventricular septal defect, pulmonary valve stenosis or atresia, displacement of the aorta to the right, and hypertrophy of right ventricle.

Tracheoesophageal fistula: An abnormal passage between the esophagus and trachea. Corrected surgically. It is frequently associated with esophageal atresia.

Translocation: The rearrangement of genetic material within the same chromosome or the transfer of a segment of one chromosome to another one. People with balanced translocations do not always manifest genetic syndromes, but may be carriers of genetic syndromes and can have children with unbalanced translocations. Can occur with any chromosomal anomaly syndrome.

Transposition of the great vessels: (Transposition of the great arteries) A congenital malformation in which the aorta arises from the right ventricle and the pulmonary artery from the left ventricle (opposite of normal), so that the venous return from the peripheral circulation is recirculated without being oxygenated in the lungs. Immediate surgical correction is needed. When this is not associated with other cardiac defects, and not corrected, it is fatal.

Tricuspid valve atresia or stenosis: A congenital cardiac condition characterized by the absence or constriction of the tricuspid valve.

Trisomy: A chromosomal abnormality characterized by one more than the normal number of chromosomes. Normally, cells contain two of each chromosome. In trisomy, cells contain three copies of a specific chromosome.

Trisomy 13: The chromosomal abnormality caused by an extra chromosome 13. Most infants do not survive beyond 6 months of life.

Trisomy 18: The chromosomal abnormality characterized by an extra copy of chromosome 18. Survival for more than a few months is rare.

Trisomy 21: See Down Syndrome.

Truncus arteriosus: See Common Truncus.

Ventricular Septal Defect: (VSD) A congenital cardiac malformation in which there are one or several openings in the ventricular septum (muscular and fibrous wall between the right and left ventricle or right and left lower chambers of the heart).

[1] Adapted from the Texas Birth Defects Monitoring Division, Texas Department of Health, http://www.tdh.state.tx.us/tbdmd/glossary.htm., Modified 2/27/01, Accessed 4/2/01.

References

Bingay E, Wilber N. Massachusetts Congenital Anomaly Surveillance Pilot Project, Interim Data Report, Office of Statistics and Evaluation, Massachusetts Department of Public Health, June 6, 1995.

Centers for Disease Control. Contribution of birth defects to infant mortality – United States 1986. MMWR, 1989; 38: 633-635.

Cragan JD, Khoury MJ. Effect of prenatal diagnosis on epidemiologic studies of birth defects. Epidemiology. November 2000, Vol. 11, No. 6: 695-699.

Edmonds L. Birth defect surveillance at the state and local level. Teratology. Vol. 56, No.1/2, July/August 1997: 2-7.

Forrester MB, Merz RD. Epidemiology of abdominal wall defects, Hawaii, 1986-1997. Teratology 1999;60:117-123.

Forrester MB, Merz RD. Impact of prenatal diagnosis and elective termination on the prevalence of selected birth defects in Hawaii. Am J Epidemiology. 1998; 148: 1206-1211.

Harris JA, James L. State-by-state cost of birth defects – 1992. Teratology. Vol. 56, No.1/2, July/August 1997: 11-16.

Hecht CA, Hook EB. Rates of Down Syndrome at live birth at one-year maternal age intervals in studies with apparent close to complete ascertainment in populations of European origin; A proposed revised rate schedule for use in genetic and prenatal screening. Am J Genetics 1996; 62:376-385.

Healthy People 2010, Volumes I and II, U.S. Department of Health and Human Services, January 2000.

Holmes LB. Congenital Malformations. In: Principles and Practices of Pediatrics 2nd Edition. Philadelphia: J.B. Lippincott Co. 1994: 277.

Kochanek KD, Hudson BC. Advanced report of final mortality statistics, 1992. Monthly Vital Statistics Report 1995; 43(6 supple.). Hyattsville (MD): National Center for Health Statistics, 1995.

March of Dimes Perinatal Data Center. StatBook: Statistics for Monitoring Maternal and Infant Health, 1997.

Massachusetts Births 1999. Bureau of Health Statistics, Research and Evaluation, Bureau of Family and Community Health, Massachusetts Department of Public Health.

Massachusetts General Laws Chapter 111, Section 67E.

Moore KL, Persaud TVN. 1993. Human Birth Defects, In: The Developing Human, 5th Edition. Philadelphia: W.B. Sounders Company.

North Carolina Birth Defects Monitoring Program Surveillance Report, October 2000.

Petrini J, Damus K, Johnston Jr. RB. Trends in infant mortality attributable to birth defects – United States, 1980-1995. MMWR. Sept. 25, 1998: 47(37); 773-778.

Sever LE, Mortensen ME. Teratology and the epidemiology of birth defects: Occupational and environmental perspectives. In: Obstetrics: Normal and Problem Pregnancies, Third Edition. New York, Churchill Livingstone, 1996.

Texas Birth Defects Registry (TBDR). Impact of including induced pregnancy terminations before 20 weeks gestation on birth defect rates. Special Report .TDH Pub No. 58-10955, Vol. 6-2. December 2000.

Trends in Infant Mortality Attributable to Birth Defects –United States, 1980-1995, MMWR September 25, 1998; 47(37):73-778.

Walker DK. Integrating birth defects surveillance in maternal and child health at the state level. Teratology. Vol. 61, No.1/2, January/February 2000: 4-8.

Yoon PW, Olney RS, Khoury MJ, Sappenfield WM, Chavez GF, Taylor D. Contribution of birth defects and genetic diseases to pediatric hospitalizations. A population-based study. Arch Pediatr Adolesc Med. 1997 Nov;151(11):1082-3